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## Genetic Services Policy Project Final Report

### Chapter 1: Introduction

The following report and recommendations reflect a body of work performed from 2004-2008 as part of the **Genetic Services Policy Project (GSPP)**, funded by the Health Resources and Services Administration, Maternal and Child Health Bureau, Genetic Services Branch.

The following primary team members contributed to the majority of the chapters:

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This work was initiated recognizing the rapid growth in the availability of medical genetic technologies that can identify persons at increased risk for acute and chronic diseases, countered by the slow pace of public policy development to guide appropriate adoption and use of these technologies. In addition, the work was guided by the understanding that diffusion of new

knowledge about human health into practice is complex, often very slow, and influenced by economic, legal, and cultural factors. The emergence of these genetic discoveries serves to add to the existing burden on health care professionals providing genetic services, who increasingly find it difficult to be reimbursed for their services and are experiencing growing demand for their expertise. The issues addressed by the project are multifaceted: how and how well does the current genetic service delivery model work? Are there alternative models that would work better? How can research relevant to genetic services be best translated into practice? And, what public policy changes do we need to get us from here to there?

To understand and describe the existing genetics health care delivery system, the GSPP investigators used existing data, compiled new data, and utilized the expertise and wisdom of many varied stakeholders to inform this work. Chapter 2 depicts the genetics health care delivery system as it exists today. While this information is useful in understanding the system across the United States, for comparison, the reader may also want to review the State Genetic Profiles that describe the genetics health care system from a state perspective rather than national perspective (<http://depts.washington.edu/genpol/GSPPproducts/stgenprofindex.htm>).

In addition, GSPP investigators and advisors considered the family experience in accessing services. This was done to better highlight potential service delivery gaps or issues as well as to consider alternative models for services. To do so, several case studies were created to give more in-depth consideration to the impact of cultural, legal, ethical, and past policy issues. For each condition for which a case study was prepared, a vignette was developed to create a context around how one might enter the medical genetics pathway for a given condition and what a family might encounter along the way. These case studies and vignettes are found in Chapter 3, and include several of the thoughtful questions raised during discussion.

In Chapter 4, we explore economic issues. First we performed a critical review of all published cost benefit or cost effectiveness papers that dealt with genetic services. We also conducted our own cost effectiveness study looking specifically at a genetic test marketed to clinicians, primarily pediatricians, that can identify individuals at risk of hearing loss if exposed to certain medications. This proved to be enlightening work not only in its conclusion but also because it demonstrated that there are, in fact, times when genetic testing may be detrimental to the health care of an individual if it causes a clinician to alter the treatment plan by using substandard treatment methods based on genetic test results.

In thinking about genetic services, it is also important to consider public perception. This work, articulated in Chapter 5, began by trying to understand how the media presents genetics information (e.g., in a positive or negative context, accurately or inaccurately) because this is where many Americans receive information about emerging technologies.

Having a grounded sense of what the existing genetics health care delivery system looks like and why, the GSPP investigators then focused on what is influencing the future and how genetic services might be provided in an alternative model of health care (Chapter 6). We reviewed which genetic services are available directly to consumers and whether they are portrayed as a clinical service. We also describe how new tests are brought to market and the factors that influence this developmental process.

Finally, Chapter 7 articulates how medical services are already being reshaped in their delivery as a direct result of globalization and other forces. Likewise, there is no reason why genetic services will or could not be provided in alternative ways given the increased use of the internet and telehealth options.

The GSPP concludes this report (Chapter 8) by offering, in a prioritized fashion, specific recommendations that are intended to increase equitable access to cost effective, appropriate genetic services for all who need them. These recommendations were developed in concert with and vetted by our GSPP Advisory Committee; however, the GSPP investigators selected and prioritized those we felt were most feasible and/or likely to result in the desired outcome.

Several additional documents are included as appendices, including a complete list of dissemination activities, an analysis of the Genetic Information Nondiscrimination Act through the advocacy coalition framework, a case study on how one system is integrating genetic services into its health care services and insurance coverage, and the changing landscape of genetic services in clinical settings.

For ease in reviewing, each Chapter is posted individually and can serve as a stand alone document, or the report can be downloaded in its entirety. It is important for readers to recognize that information presented here reflects the most accurate and current information available at the time the work was being conducted; they are encouraged to note the date stamp for each section. We sincerely hope that this collective body of work will enlighten readers and stimulate much discussion.

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