LTG Associates, Inc.

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Genetics Literacy Project

Literature and Materials Review: A Working Report

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Literature and Materials Review: A Working Report

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Produced by LTG Associates, Inc., in support of efforts to improve genetics literacy under a Cooperative Agreement between the Genetic Services Branch, Division for Children with Special Health Care Needs, Maternal and Child Health Bureau, Health Resources and Services Administration, and the National March of Dimes Birth Defects Foundation.

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Executive Summary

Scientific and medical breakthroughs in human genetics and DNA-associated technologies are having an increasing impact on the ways in which conditions and disorders are diagnosed and subsequently treated. The growth in biomedical research on the specific genetic bases for certain disorders and conditions may soon lead to targeted gene-based protocols and therapies, from sampling for the identification of predictive genetic traits to the use of gene-splicing technologies in direct treatment. However, as is commonly the case, the communication and dissemination of knowledge to the public about these breakthroughs, the promise they hold and their potential limitations, has not matched the rapid pace of scientific development. As a result, many people do not fully understand the relevance of these advances to their lives. Considerable care, therefore, must be taken to ensure that such people are well informed about the implications of biomedical research for themselves and their communities.

This document reports the results of the literature and materials review conducted by senior staff at LTG Associates, Inc., as the initial step in research toward the development of effective culturally appropriate outreach to promote genetics literacy. It is produced under contract to the March of Dimes Birth Defects Foundation, in support of the Cooperative Agreement between the Genetic Services Branch (GSB) of the Maternal and Child Health Bureau (MCHB/HRSA), and the March of Dimes, for the development of the Consumer Network for Genetic Resources and Services Information. The research process itself is ongoing and involves a process of discovery that benefits from the expertise of all the project partners to the Cooperative Agreement.

The report centers on three guiding questions that are central to understanding how individuals and families receive information about genetics and utilize this information as part of their decisions regarding health and health care. The questions are: what kinds of information regarding genetics and health currently exist; when do people encounter information about genetics and health care; and, how do people find information on health care and genetic health care in particular? These questions are starting points for the research in support of developing effective genetics literacy outreach. The literature and materials review is intended to gather available information to address these questions and to refine them as guides for further research.

This report is a culmination of a four month process of collecting, cataloguing, and reviewing a considerably diverse body of data and information sources including: professional journals; books; magazines and newspapers; websites

and on-line magazines; conference proceedings; curricular materials; and, ephemera. The collection process also drew upon the resources of partner groups and other individuals involved in the project. The review of literature and materials was processual. The method used, described as "the constant comparative method," represents an adaptation of methods used for analyzing qualitative data. The Research Associates and the Project Director engaged in a constant iterative process throughout the reading and review of literature and materials. Three coding phases were utilized to refine the team's understanding of information production and dissemination: various actors produce information about genetics and health care; that information is disseminated through multiple forms of media; and, consumers and the general public acquire and give meaning to that information through the health seeking behavior of individuals and families.

In addition, producers may disseminate information that is relevant for several of the above categories. For example, developing new standards of clinical care for amniocentesis incorporates elements of genomic research, the provision of medical services, and the formulation of government policy. Ethical concerns associated with decisions about pregnancy, and issues related to raising a child with a genetic disorder must also be taken into account. While closely linked, the categories used here capture major differences that will be important for developing effective outreach.

The first guiding question is discussed in terms of five categories of information production and dissemination: Primary Genomic Research and Biotechnology; Medical Provision and Treatment; Social Science and Policy Analysis; Community Education and Outreach; and, Science Journalism. Information about genetics is ubiquitous. Consequently, these five categories overlap thematically. While closely linked, they capture major differences that will be important for developing effective outreach. The second question considers when and under what life circumstances people encounter this information on genetics and health. There are structural and cultural forces that influence when and under what circumstance individuals and families meaningfully engage with this information. In addition, there are specific life events that serve as important milestones for the development of effective outreach to promote genetics literacy related to: reproductive health; early childhood and adolescence; young adulthood; and, aging. The third question explores the way in which people seek and find information on genetics and health. Lessons from consumer research suggest that few systematic efforts have been made to examine the health seeking behavior of individuals and families specifically in instances when choices arise regarding genetics based health issues. This final question provides a framework for addressing the gaps in knowledge that will drive the future phases of research into decision making among current and emergent consumers.

Issues of communication, cultural appropriateness, and timing are raised which must be considered for the development of effective outreach for the promotion of genetics literacy. Awareness of the factors that contribute to engagement with this information will provide guidance for the development and application of the genetics literacy toolkit by highlighting the life events and processes that encourage people to learn about the relationship between genetics and health.

The issues detailed in this report provide significant lessons learned which also raise important questions for the ongoing research process in support of the development of effective genetics education outreach. They reflect the three central questions that guided the research to this stage, and they extend our understanding of the current state of information production and dissemination regarding genetics and health. These questions will continue to guide the research, and they have raised new and interesting issues that must be considered as the research evolves. These include, but are not limited to:

- What does the general public know and understand about the relationship between genetics and health care?
- Through what channels does the general public obtain information?
- What is the minimum level of knowledge that the general public requires in order to make informed decisions about their health as new developments in genomic research and biotechnology change the biomedical view of health and the provision of medical care?
- And, how can this information be transmitted most effectively?

As the research evolves, these questions will be considered within a variety of populations and are expected to be refined and restated. As discovery continues, the materials and review process will be ongoing as a central component of continued research.

As support documents, two appendices are included with this report. Appendix 1 provides a glossary of key scientific and biomedical terms that are used in the narrative of this report, provided as a guide for the lay reader. Appendix 2 provides a listing of Internet and World Wide Web (WWW) resources, including a description of those utilized in the report, and two sections detailing other web portals of interest to the reader.

Introduction

Scientific and medical breakthroughs in human genetics and DNA-associated technologies are having an increasing impact on the ways in which disorders and conditions are diagnosed and subsequently treated. The growth in biomedical research on the specific genetic bases for certain disorders and conditions may soon lead to targeted gene-based protocols and therapies, from sampling for the identification of predictive genetic traits to the use of gene-splicing technologies in direct treatment. However, as is commonly the case, the communication and dissemination of knowledge to the public about these breakthroughs, the promise they hold, and their potential limitations, has not matched the rapid pace of scientific development. As a result, many people do not fully understand the relevance of these advances to their lives and some fear the misuse of the information and the technologies. Considerable care, therefore, must be taken to ensure that such people are well informed about the implications of biomedical research for themselves and their communities.

Although the data in biomedical research are largely gathered from specific populations, these groups are not well informed about the results or implications of this research for the health of their communities. Genetics literacy should be a goal in all communities, whether defined by race or ethnicity, geographic region, language, or other social, physical, or cultural features. In addition, while the research has been conducted on human subjects, many individuals and communities believe that they have not fully benefited in an equitable way from the findings. A combination of a lack of perceived benefit, a lack of genetics literacy, and perceptions of inequity, add to the existing highly charged climate around these issues. This is a special concern for communities that have historically been underserved by the health care system and who have been largely disenfranchised from the political and economic sectors. Any lack of openness in the relationships among these communities, the federal government, and the biomedical world can only exacerbate the consequences of the lack of genetics literacy. In fact, these populations that could perhaps most benefit from advances in genetic medicine may become more vulnerable, should they become further distanced from the knowledge necessary to make informed choices as consumers of new forms of health care.

The most significant challenge to the successful development of communication and education strategies will be the sensitive and confidential nature of genetic information and the respective individual and group sensibilities. Biomedical research is not value-free. Interpretations of the research designs for data collection and the potential uses of information on human genetics are bound tightly to multiple culturally informed conceptions of health, reproduction, and

heredity. For biomedical research, and specifically molecular biology and human genetics, to realize its full benefit in the improvement of the health of all segments of American society, effective genetics literacy campaigns must relate to the cultural and medical perspectives of diverse consumer communities. These culturally diverse perspectives will facilitate the exchange of ideas and information between the biomedical community and the people who will experience the direct impact of the rapidly evolving advances in genetic-based medicine and the resulting changes in health care.

This document reports the results of the initial literature and materials review conducted by senior staff at LTG Associates, Inc., as the initial step in research toward the development of effective culturally appropriate outreach to promote genetics literacy. It is produced under contract to the March of Dimes Birth Defects Foundation, in support of the Cooperative Agreement between the Genetic Services Branch (GSB) of the Maternal and Child Health Bureau (MCHB/HRSA) and the March of Dimes, for the development of the Consumer Network for Genetic Resources and Services Information. The research itself is ongoing and involves a process of discovery that benefits from the expertise of all the project partners to the Cooperative Agreement.

The report centers on three guiding questions that are central to understanding how individuals and families receive information about genetics and utilize this information as part of their decisions regarding health and health care. The questions are:

Question 1. What kinds of information regarding genetics and health currently exist; who is generating and transmitting information on genetics and health care; what modes of transmission are used; and what is the general content of the information?

Question 2: When do people encounter information about genetics and health care?

Question 3: How do people find information on health care and genetic health care in particular?

These questions are starting points for the research in support of developing effective genetics literacy outreach. The literature and materials review is intended to gather available information to address these questions and to refine them as guides for further research.

The report is structured as follows. The methodology section provides a full description of the collection of literature and materials for review, the systematic

methodology utilized for analysis and review, and the guiding principles in the search process. The body of the report is then organized around the three guiding questions, with each considered as separate data and analysis sections.

For the first question, the literature and materials are organized into five categories of information production and dissemination: Primary Genomic Research and Biotechnology; Medical Provision and Treatment; Social Science and Policy Analysis; Community Education and Outreach Initiatives; and, Science Journalism. In a summary the interrelationship between these categories of information and its impact on the dissemination of information to the public is discussed.

For the second question, the discussion begins with considering the influence of structural and cultural forces on access to and encounters with information about genetics and health care. Then, a life cycle approach is utilized to discuss different points at which an individual or family may become meaningfully engaged with information about genetics relevant to their health. A summary discussion of the importance of understanding differences in engagement with genetics information by individuals and families for the development of effective outreach is then provided.

For the third question, there is little current information on how consumers actually seek and find information on genetics and health. In addition, while there are well known models of health seeking behavior, they have not been applied specifically to understanding the decision making processes by individuals and families regarding genetics based health care issues. Materials are presented in this section from current consumer research and extant models of health seeking behaviors of individuals and families. Special care is given to considering how new protocols and treatments stemming from developments in genetic-based medicine will essentially reframe health seeking behaviors, many of which are bound closely with cultural conceptions of illness and health.

The report concludes with a discussion of the lessons learned and questions raised in this literature and materials review process. Most notably, issues of communication, cultural appropriateness, and timing are raised which must be considered for the development of effective outreach for the promotion of genetic literacy. This review offers valuable lessons for guiding future steps in the research process and for developing effective outreach messages and strategies.

Two appendices are also included with this report. Appendix 1 provides a glossary of key scientific and biomedical terms that are used in the narrative of this report, provided as a guide for the lay reader. Appendix 2 provides a listing of Internet and World Wide Web (WWW) resources, including a description of

those utilized in the report, and two sections detailing other web portals of interest to the reader.

Methodology

This report on the literature and materials review is a culmination of a four month process of collecting, cataloguing, and reviewing a considerably diverse body of data and information sources including: professional journals; books; magazines and newspapers; websites and on-line magazines; conference proceedings; curricular materials; and, ephemera. The collection process did not include relevant materials from non-print media such as radio, television, and film.

Collection of Materials

The early stages of collection relied on keyword and keyword combination searches in a variety of media: websites and catalogues; web based journals and newspapers; science magazines and books targeted for the lay audience; peer reviewed journals; and, government reports pulled from official government websites. Primary search engines included ERIC, Medline, Bioethicsline, Histline, Popline, as well as general web search engines such as Altavista and Yahoo. Searches used key words, and key word combinations of the terms beginning with: genetics; genes; genomics; culture; ethnic; ethnicity; ethics; bioethics; morality; education; eugenics; and, behavior. All searches were downloaded in either text format or referencing software format. Medline searches were then uploaded into Endnote 4.0. These references were fully annotated, complete with abstracts and keywords. Bioethicsline, Histline, and Popline references do not contain full abstracts but do have subject headings. Text only references were uploaded manually into Endnote 4.0 primarily using cut and paste procedures. Abstracts and subject headings allow for effective sorting of the literature along categorical axes during this and subsequent research phases.

Full text articles used for reading and review were collected during several visits to the National Library of Medicine and the University of Maryland College Park. These full text articles are archived as hard copy at the LTG Library in the LTG Offices at Takoma Park.

A wide variety of materials were also collected from Internet sources. Given the close relationships between Internet information technologies and the bioinformatics associated with genomic research, research staff collected a substantial amount of portal web pages, reports, and full text articles. One way to categorize websites is to focus on type of web host such as: federal agencies; state and regional agencies; professional agencies; university based institutes or programs; private not-for-profit organizations; and, private industry and research facilities. This categorization was useful for research oriented reading and

review. However, these nested websites are linked together in multiple ways, and any consumer searching for information can follow a variety of complex paths to information. Therefore, this categorization cannot unpack the complexity of how consumers may acquire information. Instead the review process distinguishes between materials based upon the specific target audience, content of information, and accessibility.

The Internet is a valuable resource for collecting government reports and policy statements, information from biotechnology, and other popular sources from news wires. Efficient downloading requires high-speed access and sufficient bandwidth. The materials collected from government and most popular sources are provided at no-cost for public use. Some Internet sources target scientists, medical professionals, and biotechnology professionals. GeneTest, for example, is a not-for-profit web resource sponsored by the National Library of Medicine of the National Institutes of Health (NIH) and MCHB that offers information about genetic counseling and genetic testing. Geneletter, a free web magazine sponsored by GeneSage, a for-profit company, is a good example of one source of well-reviewed information on developments related to genetics and health. Sites sponsored by Medscape, Biomednet, The Scientist, Nature Genetics, and other similar resources require users to subscribe at no cost. The subscription process acts as a kind of firewall or filter that encourages users to stay within the site. These sites provide extensive information, much of which may be appropriate for broader public consumption. Materials from websites sponsored by the Department of Energy (DOE) and the National Institutes of Health (NIH) provide valuable background information. These were supplemented by full-length studies that place the Human Genome Project (HGP) and related biotechnologies in historical context.

Keyword searches on major search engines such as Yahoo or Altavista usually resulted in too many hits to be of value. However, the process reveals the kinds of information and discussions related to genetics and health care both within and outside the mainstream medical and scientific community. For example, a search using the term “eugenics” links to high confidence hits for a number of extremist political groups and organizations. A term such as genomics, on the other hand, yields hits for biotechnology firms, federal websites, universities, and others. As a result, the initiate to Internet-based searches for information related to genetics and health will be presented with a spectrum of sites from mainstream and non-mainstream sources, many of which are not reviewed for scientific or medical accuracy. In addition, there are no clear markers for the consumer to use to determine the validity of information accessed.

The collection process also drew upon the resources of partner groups and other individuals involved in the project. This provided access to: non-published proceedings from recent professional conferences; research and funding

proposals; materials designed for professional education; consumer-oriented pamphlets; and, background literature on recent and ongoing projects related to genetics and health. These materials provided background information on how various actors have developed projects and programs related to genetics.

Analysis: The Constant Comparative Method

The review of literature and materials was processual. The method used, described as "the constant comparative method," represents an adaptation of methods used for analyzing qualitative data. The Research Associates and the Project Director engaged in a constant iterative process throughout the reading and review of literature and materials. Throughout this process, the Research Associates conducted the initial readings. The Project Director in turn suggested new areas for further research, and helped guide the process of creating taxonomies and relating the analyses of different types of materials to the goals of the project. In practical terms, this meant developing a process in which the Research Associates and Project Director exchanged views and analyses as a team, thus ensuring consistency of perspective and terminology.

This iterative and interactive process relied on a series of steps that began with open coding. In this first step, the Research Associates relied on text-searching within documents and in Endnote 4.0. Documents were categorized and labeled using a number of criteria including but not limited to: biotechnology; primary medical research; professional medical education initiatives; social science and policy analyses; and, popular and journalistic accounts. The categories represented the first initial sort of the materials and reflect the general process by which information about genetics is produced and then transformed and disseminated.

Following the initial sort, the Research Associates and Project Director collectively engaged in a process of axial coding. During axial coding, documents from the various categories are subject to more intensive review. At this stage the team focused on three axes or dimensions. The first axis pertains to the purpose of the document, e.g., reporting new research findings, provider or consumer education, or analysis of trends in technology or society. The second axis pertains to the target audience. The third axis pertains to the media through which the document is disseminated. During the axial coding process, the researchers were able to link documents across categories and capture the ways in which documents can serve multiple purposes.

It is important to note that the team has moved constantly back and forth between both forms of coding, especially as new materials were collected and they refined the categories and sub-categories being used.

The next phase in the process is selected coding. This portion of the coding proved particularly important for those documents that pertain to issues of ethics, policy, social scientific analyses, and social criticism. The selected coding process focused on identifying trends, concerns, and styles of argumentation that are commonly found in the literature across the categories developed during the open and axial coding process. For example, one trend is the tendency to speak optimistically about the "revolutionary" developments in genetics research. There is also a significant trend of pessimism or concern regarding the unfettered use of new technologies. During this selected coding process, the team analyzed these trends within the broader system of biotechnology, biomedicine, and American culture, and then re-connected documents in a holistic fashion.

This final phase became particularly important for refining the team's understanding of three stages in the process: various actors produce information about genetics and health care; that information is disseminated through multiple forms of media; and, consumers and the general public acquire and give meaning to that information in a variety of ways.

Guiding Principles in the Search Process

Advances in genetic research have had a significant and broad impact in the scientific, biotechnology, medical, social science, and public policy arenas. In addition, it has generated wide debate and interest in media and popular culture. Consequently, there are several ways in which information related to the advances in genetics research may be categorized. This review has relied on two complementary systems of classification. The first classification, referred to as the process classification, is oriented toward information generation and dissemination. It is derived from three guiding questions about how people interact with genetics and health care. First, what types of actors are currently producing and disseminating information regarding genetic-based health care? Second, when and how do people, if ever, become meaningfully engaged with genetics and health care beyond a superficial scanning of the media? And third, how do people actively find information about genetics, medicine, and health as part of medical decision making processes.

The second classification, referred to as a content classification, was developed in order to effectively archive and access the variety of materials collected for purposes of review. Materials were oriented along three different axes. The first focused on the target audiences, including but not limited to: professionals and experts; media niche markets such as current consumers, emerging consumers, and the general public; potential biotechnology stakeholders and investors; and, policy makers and government officials. The second axis focuses on the form the materials take, including: conference proceedings and presentations; peer

reviewed journal articles; popular magazines; government reports; reportage in the general press; websites; online magazines; and, education program materials. The third focuses on the nature of the content in the materials. This axis varies considerably, but as examples it includes: a discussion of a particular condition or disorder; a technological innovation; an advance in medical treatment; a moral and ethical discussion; or, an analysis of social, economic and political implications.

These categorizations have been integrated to produce a detailed taxonomy of the literature that will in turn assist in framing research issues during the subsequent phases. The category of producers and disseminators structures the first section of the report in consideration of the first guiding question. Issues of target audience, form, and content are then integrated into each subsection. The first section then concludes with a critical discussion of the complex ways in which producers and disseminators overlap with one another. The next two sections focus on consumers' engagement with genetics and health care as the other two guiding questions are discussed. In sum, section one describes what groups of actors are involved in producing and disseminating information, and the latter two provide a framework for understanding how consumers may or may not become engaged in issues related to genetics and health care. Of course, the issue of engagement is to be considered as a process of discovery in subsequent research phases that focus on consumers and the general public. This framework represents an initial sort of the materials currently available and it is intended to produce lessons learned from the current phase and to focus questions for subsequent research.

Sources of Information on Genetics and Health

Question 1. What kinds of information regarding genetics and health currently exist? Who is generating and transmitting information on genetics and health care; what modes of transmission are used; and what is the general content of the information?

Information regarding genetics and health is produced and disseminated through a combination of sources, some acting in tandem, others singularly. As information is produced through biotechnology and biomedical research, it is then disseminated and captured through a variety of media outward toward the public. The aim of this introductory section is to provide a general framework for understanding the complex interactions and feedback mechanisms that are driving innovations in biotechnology, primary genomic and medical research, and the resulting dissemination of information regarding genetics and health care.

The literature and materials are organized into five categories of information production: Primary Genomic Research and Biotechnology; Medical Provision and Treatment; Social Science and Policy Analysis; Community Education and Outreach Initiatives; and, Science Journalism. The major categories of information production represent one heuristic device that imposes order on a remarkably diverse collection of materials. Each category represents a collection of actors--individuals, private companies, public entities, and associations--who produce relevant information on genetics and health care. Tracing how actors filter, funnel, and package that information is a considerable task and this review offers a general outline of these processes.

The categories are useful for organizing the wide scope of materials and literature collected thus far. Nevertheless, any categorization also imposes order on a set of activities and practices that overlap and intertwine in complex ways. New developments in primary genomic research and biotechnology feed into each other. Technological innovations in gene sequencing and mapping for example, have had a profound effect on the speed in which new candidate genes for various disorders are discovered. The discovery of candidate genes in turn raises issues of patenting and intellectual property, information sharing that may lead to new kinds of testing, and ethical debates about the ownership of genes or pieces of DNA. Research into testing and new technologies may generate new kinds of hypotheses that feed back into primary genomic research. In short, the integration of developments in bioinformatics, new technologies, and primary research is very strong.

It is also difficult to separate these developments from the biomedical mission to develop new understandings of the etiology of disorders, and the promise those understandings generate for improving health care. Proteomics, pharmacogenomics, gene transfer therapy, pre-implantation diagnosis, amniocentesis, and stem cell and embryo research are all areas in which technology, medical treatment, and ethical issues are intertwined. In addition, all of these developments take place within a wider social and cultural context in which popular notions of heredity are intertwined with folk conceptions of social and environmental influence. The nature/nurture controversy, however inadequately characterized in the popular press, is another theme which serves as a background to how information is received by the public. How information is disseminated among groups of experts and how it ultimately filters into the popular consciousness is central to understanding access to information about genetics and health as a whole.

These categories of information production are designed to reflect how information regarding new developments and discoveries are initially reported and then repackaged and disseminated to wider audiences, whether those audiences are the medical professionals, members of the biotechnology and pharmaceutical industries, or, for the purposes of this report, current and emergent consumers, and the general public. In discussing each of these categories, three main themes are covered that integrate the different stakeholders into the overall process referred of generating and disseminating information on genetics and health care. The first theme relates to describing the major technological and medical developments associated with genomic research. The second theme relates to understanding the ways in which these developments engender ethical, legal, and social issues. The third theme emphasizes how the first two themes and issues are translated, often altered, at times compromised, and ultimately received by diverse audiences as information is disseminated.

It is important to note here that for purposes of this report a sharp distinction is not imposed between the public sector and private sector initiatives. Both pubic agencies and private sector firms support the Human Genome Project (HGP). The DOE Human Genome Program and the NIH National Human Genome Research Institute (NHGRI) provide funding for primary genomic research conducted as part of the HGP. They also support primary research into the social, ethical, and legal implications of this effort. Both the DOE and NIH have developed cooperative relationships with the private sector, including for-profit firms such as Celera Genomics. Private not-for-profit agencies, foundations, and professional associations have also been closely involved in the HGP. It is therefore impractical as a guide for this research to impose a sharp demarcation between types of initiatives since information production and dissemination is through cooperative and integrated channels.

In addition to the private/public sector linkages created by the HGP, a variety of government agencies develop and disseminate valuable information and educational materials regarding genetics and health care. These include, but are not limited to: MCHB, and its Genetic Services Branch (GSB); the Centers for Disease Control and Prevention (CDC); state and local health departments; and, state level genetics services organizations. These government agencies all have different mandates and functions. Some, such as CDC, provide information to the public via their websites and publications. Others, such as GSB, use funding mechanisms for developing targeted programs related to health and genetics. In addition, there have been concerted efforts to coordinate the activities of different federal agencies, as reflected in the cooperative efforts of the Agency for HealthCare Research and Quality (AHRQ), CDC, HRSA, and NIH (see "The Memorandum of Understanding to Enhance the Translation of Genetics into Health Policy and Practice"). One example of their coordinated effort has been the three annual Public Health and Genetics Conferences convened by the Association of State and Territorial Health Officials (ASTHO), CDC, HRSA, and NIH (Centers for Disease Control and Prevention 1998).

It is emphasized here that the complex functional and funding linkages between the public and private sectors make it difficult to make firm statements that relate the type, content, and quality of information to particular sectoral distinctions. Instead, the categories that have been created attempt to capture the larger macro-processes by which the raw data of primary genetics research is translated into information valuable to experts in related fields, and then is ultimately transformed and disseminated as knowledge that may help consumers make informed decisions about their health care.

Primary Genomic Research and Biotechnology

This category captures information production from a diverse collection of primary research efforts and the development of associated technologies. This includes but is not limited to: the biotechnology procedures that have made it possible to manipulate genetic material using cloning vectors such as bacteria or viruses; research on techniques for DNA sequencing, cloning, and protein transcription such as gel electrophoresis and sequence tagging; and, research in molecular and cell biology, protenomics, and developmental biology such as stem cell and pre-embryo research. Research on the comparative genomics of non-human organisms such as *C.elegans*, *Drosophila*, and mice, is also included. Reporting of research within science and industry is largely intended for scientific specialists and professionals and is therefore of a highly technical nature. It is not intended for direct consumption by the general public or even current consumers of genetics services. As a result, the review of this category will focus on the types of information that is accessible to the public.

Through their grant mechanisms, the DOE and NIH represent two principle drivers of primary genetics research in both the public and private sector. The Internet is one of the primary channels for disseminating technical information. Virtually every research institute and biotechnology firm that focuses on primary research maintains virtual databases and websites for ease of access and rapid communication. The websites maintained by DOE (www.ornl.gov/hgmis), and NIH (www.nhgri.nih.gov) archive and channel the latest research findings for the community of researchers. To some extent they also provide information to the public about the overall scope and potential implications of the projects. The DOE website devoted to the HGP for example, integrates the wide variety of materials that capture the scope and potential implications of research associated with the HGP. These include links to news sources, educational resources for teachers and students, fact sheets, a section on the ethical, legal and social issues, and general descriptions of gene testing, pharmacogenomics, gene therapy, genetics counseling, and genetic disorders and conditions.

The research section of the DOE website offers descriptions of and hyperlinks to sites where sequencing information in the public domain are stored. These focus on bioinformatics, functional and comparative genomics, sequencing and sequencing technologies, mapping, and virtual library genetics. The same section also contains links to information about funding opportunities, and information on U.S. and international research sites. The Researcher Resources section of the NIH NHGRI website (www.nhgri.nih.gov/Data) offers a second example of access to primary research databases including the Genome Data Base, The European Bioinformatics Institute, The Eukaryotic Promoter Database, or the Databank of Secondary Structures of Proteins, and to university based and private genome centers such as the Institute for Genomic Research, University of California at Berkeley Drosophila Genome Center, or the Whitehead Institute. The DOE and NIH websites offer different channels for visitors, depending on their backgrounds and interests. Lay visitors are encouraged to access certain portions of the site, while other portions are clearly designated for experts and researchers.

Domestic and international biotechnology firms provide thousands of websites, reports, publications, and promotional materials on the Internet. Access to specific biotech company websites requires knowing the firm's name, its URL, or the relevant keywords. Without knowledge of the requisite technical keywords, gaining access to these sites would be time consuming. Therefore, consumers are less likely to access this information without a targeted search. For industry insiders however, there are Internet companies such as Resource Informagen (www.informagen.com), that provide free directory listings for firms and businesses involved in biotechnology.

It is clear that the websites and materials offered by biotechnology firms are directed towards the business and research communities and not directly to consumers. Nevertheless, company descriptions of their corporate mission, product development, and financial standing are all placed within the larger context of the HGP, medicine and genetics, pharmacogenomics, and other genetics related subjects. Furthermore, many of the marketing divisions of such firms have used the Internet and conventional electronic dissemination methods such as press releases to promote their findings and products. Some of the information filters through to the business and technology sections of newspapers and other media outlets through science writers and other journalists. Information from such private sector sources is not necessarily differentiated by the public from basic research findings in the public sector and academic sources. As a result public perception of developments in genetics relevant to health combine information from all of these sources.

Medical Provision and Treatment

Information on genetics and health care is generated and transmitted by those in the medical profession engaged in research or the provision of health care. The review of the literature demonstrates that the modes through which this information is channeled are incredibly diverse, and depend, in part, on the intended audience of the information. Broadly speaking, this information is intended either for health care researchers, providers, or their patients as consumers of health care services.

Health care providers and researchers share information on genetics and health care through the use of web-based media, conferences, and print media including professional journals and text. Information produced by and intended for a professional audience comes in a number of forms. Highly technical, clinical reports on genetics research developments and their health care applications constitute one form of information. Other information is presented in a format that emphasizes the need for increasing genetics competency among professionals. This genre of literature is predicated on the belief that advances in genomic research will fundamentally change the nature of health care, including consumer health needs and expectations (Billings 2000a). It includes: educational material and course curricula; professional commentary; and, published guidelines for diagnosis and treatment of genetic-based conditions. As a whole, this information is useful for understanding up-to-date research in the field of genetics and health care, the issues currently targeted by medical researchers and providers for increased genetics competency, and how this information may influence the education of consumers.

Consumers may receive information produced in the fields of health care and service provision by visiting websites or reading professional journals. However,

few systematic attempts to specifically educate consumers have been made by health care researchers and providers. The greater emphasis on provider education is a result of the traditionally hierarchical model of education that guides professional discourse on genetics and health care. According to this model, consumers rely on health care providers for education, so educating providers on the latest developments in genetics will result directly in increased genetics literacy among consumers.

A body of research has suggested that consumer-provider interactions are a principal means by which health care information is disseminated to consumers. This literature review at this stage has not focused on the nature of information transmitted to consumers by health care providers through clinical office visits. However, the review suggests that consumer-provider interaction built on the hierarchical model of education is the primary determinant of consumer knowledge about genetics and health care at this point in time. Because this is one of the primary means by which providers communicate information on genetics and health care to consumers, it is vital to further explore this mode of communication in the future phases of the research.

Numerous medical specialties make up the category of health care research and provision, each adding a different dimension to the extant information on genetics and health care. The following overview summarizes the topical areas of greatest concern within each medical specialty and suggests reasons why understanding such information will be important for the Project and its partners.

Pediatrics

People working in the field of pediatrics are intimately involved in the production and dissemination of information on genetics and health care. This engagement with genetics emerged from early efforts aimed at treating childhood disorders such as cystic fibrosis and phenylketonuria (PKU). Much of the pediatrics literature focuses on "phenotypic prevention," or medical interventions designed to forestall and/or treat the physical expression of genetic disorders (see Juengst 1995b). For example, this phenotypic prevention model guides discourse on: the role of folic acid in preventing neural tube defects (1993; 1999); the treatment of tuberculosis (1994); the importance of psychological support in health maintenance of genetic disorders (Carey 1992); the treatment of cystic fibrosis (MacLusky 1993); and, the establishment of guidelines for the management and treatment of PKU (Seashore, et al. 1999; Wappner, et al. 1999). Understanding the pediatric emphasis on phenotypic prevention provides insight into public health-driven programs designed to prevent or reduce morbidity and mortality among those with specific genetic profiles (Khoury, et al. 2000, p.6).

Those working in the field of pediatrics produce highly technical information that reports developments in genetics research with practical implications for health

care provision. Examples include: reports on screening for genetic markers of congenital adrenal hyperplasia (CAH) (Ferenczi, et al. 1999); the role of genes in growth (Martorell, et al. 1989); the current state of knowledge on genetic disorders such as trinucleotide repeat disorders (O'Donnell and Zoghbi 1995); microcephaly (Opitz and Holt 1990); Rett syndrome (Percy 2000); renal disease (Saborio and Scheinman 1998); pediatric cardiovascular disease (Strauss and Johnson 1996); and, the influence of genetics on child behavior problems (Porges, et al. 1992). While much of this information is highly technical and intended for providers and researchers, it affects consumer engagement with genetics services by influencing the information and services that providers have at their disposal and offer to consumers as they make decisions about their health.

Much of the pediatric information, like information in all medical fields, is explicitly designed to assist providers in meeting the anticipated demand for genetics services. This category of information includes reports on the significance of advances in molecular biology to pediatric practice and therapy (Gansler 1991; McCabe 1999; Mullis and Wagner 1995; Potter and Tarleton 1998; Raffel 1997; Smith 1997) and educational primers on molecular biology (Demetrick 1994; Diamond and Eichler 1999; McCabe 1997; O'Leary and Wright 1991). Included in this category is information on the relationship between genetics and health that focuses on diagnostic approaches to genetic disorders (Clarke 1992; Peeden and Noe 1992), and the development and evaluation of genetic tests, such as screening for maternal serum alpha-fetoprotein (1991), HIV (Avila, et al. 2000), and diabetes (Guazzarotti, et al. 1999).

Standards and guidelines for genetic screening and prenatal diagnosis established by the American Academy of Pediatrics (AAP), Committee on Genetics is part of the larger effort to educate health care providers (1992) (1993), (1994b). The AAP Committee on Genetics also disseminates information that establishes standards for the health supervision of children with genetic disorders such as: Down syndrome (1994a); Turner syndrome (1995c); neurofibromatosis (1995b); achondroplasia (1995a); Marfan syndrome (1996b); sickle-cell diseases (1996c); and, fragile-X syndrome (1996a). Part of this effort to educate providers of pediatric care includes evaluations of genetics services provided by pediatricians (DeClue and Schocken 1991; Liacouras and Shamir 1997).

Part of the pediatrics literature raises awareness of the ethical challenges genetics technology presents to pediatricians (Shapiro 1993; Van Allen 1997), such as, but not limited to: the implications of testing children for cancer risk (Kodish 1999; Laxova 1999); the future importance of offering genetics counseling to consumers (Phadke and Pandey 1999); and, the new developments in gene therapy (Ledley 1996). Although this information is not

intended for the direct education of consumers, it contributes to the interactions between providers and consumers, affecting the way in which information and services are transmitted.

Overall, this educational material is vital to the development of a toolkit for improving genetics literacy, particularly when considering current consumers. However, the degree to which such materials, standards, and guidelines are based upon an awareness of cultural difference among consumers must be taken into account. Standards established for the treatment of a "generic" consumer may provide insight into current guidelines for genetic screening and treatment, but may not adequately emphasize culturally appropriate approaches to meeting the genetic-based health care needs of underserved populations. Engaging this literature also provides an avenue for exploring the social, legal, and ethical issues associated with genetic testing.

Obstetrics-Gynecology

The field of Obstetrics-Gynecology (OBGYN) has a long historical engagement with genetics and health care issues due to advances in prenatal diagnosis and testing. Amniocentesis, used in the 1950s to test for maternal-fetal blood group incompatibility, has since become a common method for assessing fetal genetic health. New prenatal diagnostic technologies have become available since the development of amniocentesis, including chorionic villus sampling (CVS), and maternal serum alpha-fetoprotein screening. Advances in assisted reproductive technologies including preimplantation diagnosis also encourage an active engagement between OBGYN specialists and genetics information.

Whereas information generated in the field of pediatrics often emphasizes phenotypic prevention, practitioners in OBGYN must confront the issue of "genotypic prevention." Genotypic prevention involves predicting the potential transmission of particular genetic material to succeeding generations, often resulting in selective abortion based on prenatal diagnosis (Juengst 1995b). Due to the ethical issues attached to genotypic prevention, understanding this distinction between phenotypic and genotypic prevention is vitally important to account for differences in ethical considerations.

A tremendous amount of OBGYN literature reports on clinical research into the accuracy, risks, and benefits of these diagnostic tests. Such emphasis on technological assessment has been criticized by those who believe that more attention must be given to the concerns and experiences of women being tested (Rapp 1999, p.32). There are vital ethical issues involved in such prenatal testing, since genotypic prevention approaches are predicated on selective abortion in the case of certain deleterious traits, which could lead to a medically defined "handicap" in a child. For many women selective abortion is not an option and many mothers and families choose to challenge the very notion of

"handicap" or "disability." This critique strongly highlights the need for more research on consumer knowledge, beliefs, and attitudes in developing culturally appropriate information and services.

As genetic testing during the prenatal phase moves from the realm of monogenic disorders to the diagnosis of polygenic disorders, some of which may not be fully expressed until adulthood, even greater interest in ethical issues and dilemmas is being generated (Milunsky and Milunsky 1997; Penticuff 1994). One such issue involves the potential practice of selective abortion in cases involving identified risk for Alzheimer's disease should markers be identified by prenatal diagnosis (Post, et al. 1992). Due to advances in genomics research and the discovery of new genetic markers for health risks, OBGYN practitioners are emphasizing the need for sensitivity and concern for consumer understanding of medical risk (Grimes and Snively 1999). One of the challenges to be faced is understanding consumer perception of risk and the underlying knowledge, beliefs, and attitudes that guide practice. In addition, there is a growing recognition of the role and importance of genetics support groups for consumers (Mackta and Weiss 1994), and parental strategies for coping with pre-existing or emergent genetic disorders (Olsen 1994). Although much of this information is targeted specifically to OBGYN providers, the content is also useful for Project partners in understanding genetic health care issues specific to OBGYN, and more generally, the ethical concerns raised therein.

Oncology

Information on genetics and health care produced and disseminated within the field of oncology highlights the rapid pace of research on the human genome, the ethical issues involved in genetic screening for cancer, and the profession's emphasis on provider education. The discovery of genes associated with heightened cancer susceptibility, such as BRCA1 and BRCA2 (associated with breast cancer), and polymorphisms of the P53 gene (associated with colorectal cancer), has raised concerns over predisposition tests (Calzone 1997). Questions are raised about such issues as: the translation of genetic predisposition into risk assessment; interactions between genetic predisposition and environment; or, the choices available to those found to have a particular genetic predisposition. These developments have encouraged debate on the ethical issues surrounding cancer screening (Vineis 1997).

Oncology literature and information is similar to that produced in other medical disciplines in a number of ways. Oncology specialists are concerned with presenting new clinical research findings to the medical community (Clericuzio and Johnson 1995), and with establishing protocols and guidelines for genetic testing (Offit, et al. 1996). Like other medical disciplines, providers in the field of oncology recognize the need to enhance their own genetics literacy and the need for new courses of study based on developments in genetics research (American

Society of Clinical Oncology 1997b; Bowles 1997). Within this discourse providers are emphasizing the need for increased sensitivity to consumer preferences regarding testing and counseling (Audrain, et al. 1998; Stopfer 2000). Information designed for oncology nurses, in particular, emphasizes the importance of being competent in communicating and discussing risk, understanding consumer concerns, and providing psychosocial support (Calzone 1997; Lea 1997; MacDonald 1997). The oncology literature is significant because it highlights the practical and ethical concerns that emerge from the rapid discoveries of genetic markers for complex or delayed onset disorders. It also demonstrates how genomic research is driving the discourse on genetics and health care and enhancing the perceived need for improving education among health care providers.

Nursing

Nursing literature emphasizes the need for improving genetics literacy among nurses in order to keep pace with the rapid advances in genomic research (Anderson, et al. 2000; Anderson 1996; Feetham 1999; Gottlieb 1998). Among health care providers, nurses have demonstrated leadership in creating genetics literacy initiatives (Monsen 1996) and are at the forefront of efforts to develop community-based public health outreach programs (Affonso, et al. 1993). According to this literature, nursing must play an important role in the face-to-face interaction with consumers of genetics services, providing proper education and counseling. However, unlike the traditionally hierarchical education models implicit in much of the primary care literature, literature in nursing highlights the need for a culturally appropriate, holistic, and dialogical approach to consumer education (Anderson 1998; Lea, et al. 1998; Loescher and Ronan 1998; Silva 1994). This information will be vital for facilitating the design of a genetics literacy toolkit that will result in culturally appropriate messages.

The emphasis in the oncology nursing literature reflects the general interests and concerns regarding genetics and health in the nursing literature as a whole. Much of the information being produced asserts that the rapid pace of discoveries in human genome research, coupled with the perceived shortage of qualified, certified genetics counselors, places increased demand on nurses to provide consumer education and counseling (Lashley 1997). It is important for Project partners to be aware of this movement towards self-education in the nursing profession, and among health care providers in general.

Considering the consumer-oriented approach of educational efforts in nursing, it is not surprising that much of the literature addresses the ethical, social, and legal dimensions of genetics and health care (Diekelmann 2000; Monsen 2000; Roth and Painter 2000). One particular focus is the concern to find ways to make the delivery of genetic health care and the handling of personal genetic information fundamentally ethical (Kirk 2000; Scanlon and Fibison 1995).

Included in this literature is information designed to educate nurses on the historical context within which genome research is embedded and the association between genetics and eugenics (Iredale 2000; Jenkins 2000). As a result, the nursing literature is similar to other medical disciplines in emphasizing provider education, but it also provides an important consumer-oriented approach to genetics literacy that underscores the need for holism, culturally appropriate education, and an historical awareness of the social context within which information is understood and genetic health care is provided.

Primary Care

Information provided by and for primary care providers, e.g., family practitioners, general practitioners, primary care physicians, include reports on clinical genetics, measurement of risk factors, and other medical research findings. However, this literature varies in terms of basic assumptions about the future of genetics based health care, falling between two ends of a spectrum.

On one end is literature based on the commonly held assumption that a revolution in genetics is at hand, requiring a greater level of knowledge and sophistication for the proper provision of an expanding number of genetics services (Fears, et al. 1999). Information at this end of the spectrum is generally optimistic about the future role of genetics in medicine, and it encourages active engagement with new genetics knowledge and developments. Similar to literature in nursing, it is based on the assumption that the shortage of genetic counselors and specialists will place a greater burden on primary care providers, and depicts a future in which providers are challenged to meet the needs of consumers looking for the latest development in genetic testing for improving health. Most of this literature is optimistic about the future role of primary care in providing consumers with accurate information (Kolb, et al. 1999). Included in this literature are: studies on the effectiveness of new teaching materials on student acquisition of genetics knowledge (Teague, et al. 1996); the type of information consumers are most likely to request from their primary care provider (Facher and Robin 2000); and alternatives to direct provider-consumer education, such as computer assisted learning tools (Green and Fost 1997).

At the other end of the spectrum are those who are less certain about the degree to which the new discoveries in genomics will revolutionize medicine. According to this view, the lack of effective treatments for those with genetic disorders, coupled with public concerns over gene therapy and testing, diminishes the potential role of genetics in health care, despite the development of new genetic diagnostics (Holtzman and Marteau 2000; Wulfsberg 2000). Providers at this end of the spectrum generally suggest that current methods of disseminating and communicating information, such as professional journals, websites, newsletters, and conferences are sufficient. In short, they do not see

the need for aggressive genetics literacy campaigns targeted toward consumers or primary care providers.

Studies on physician attitudes towards genetics and their role in the provision of genetics services tend to reinforce the former perspective. That is, although genetic health care constitutes a fraction of the services and information currently provided in primary care, physicians view it as a future priority. These studies indicate that primary care physicians consider themselves to be on the front-line of the growing demand for information on genetics and health care. They also suggest that most primary care physicians recognize that they must play a greater role in genetic health care, but currently lack the confidence and genetics literacy to do so (Emery, et al. 1999; Friedman, et al. 1997; Watson, et al. 1999). These studies provide insights into the practical and methodological challenges associated with efforts to improve genetics literacy among primary care providers that effect consumer education (Geller and Holtzman 1991). More significantly, they provide valuable information on the attitudes and practices of primary care providers, and how they might influence consumer education and decision making processes.

For example, one study suggests how provider perceptions of disability may influence the types of genetics information and services offered to consumers (Kirschner, et al. 2000). Another study found that most physicians assume the responsibility of genetics counseling, typically providing referrals to genetics specialists only after consumers expressed a deliberate interest in the service (Hayflick, et al. 1998). This finding reflects a broader theme underlying much of the primary care literature. In short, primary care providers assume the role of gatekeepers to the world of genetics and health care, providing primary education for consumers according to a prescriptive or directive approach to care. There have been efforts, however, to create continuing education material for primary health care providers (Fine and Koblenz 1994).

Genetic Counseling

The directive nature of interactions between primary care providers and consumers contrasts sharply with the nondirective imperative espoused in the literature produced by and for genetic counselors (Dialey, et al. 1995; Yarborough, et al. 1989). Since its inception as a formal discipline in 1969, genetic counseling has emphasized the importance of communicating value-neutral information to consumers. This emphasis is based on a conscious attempt to support the health care decisions of autonomous consumers, which stems, at least in part, from the desire to distance the discipline from directive eugenics policies of the early 20th Century. Much of the literature focuses on the practical and ethical problems associated with this philosophy and the inherent contradiction of offering, "a value-charged technology in a value neutral manner." (Rapp 1999:59). It also highlights the philosophical divide between the

directive approach of primary care physicians and the nondirective model of genetic counseling. The former emphasizes improved health outcomes, while the latter privileges informed and autonomous individual decision making.

Like primary care physicians and nurses, genetic counselors perceive themselves as being on the front-lines of genetics and health care, confronting the challenges of improving genetics literacy on a daily basis. Literature in this field is particularly useful because it covers an expansive array of practical, methodological, and ethical issues surrounding the education of consumers. The literature reflects the intent to communicate information on genetics to consumers holistically, in light of the complex ethical, social, and legal issues surrounding the discipline (Bartels, et al. 1993; Rothstein 1993). Among the practical and ethical issues confronted by genetic counselors are: problems of communicating risk; balancing the consumer's need for simple information with the non-directive approach; the importance of informed consent; assuring confidentiality; and, minimizing the potential for discrimination (Uhlmann, et al. 1996). Guidelines for practice and core competency requirements established by the American Board of Genetic Counseling are particularly informative (Fiddler, et al. 1996; Fine, et al. 2000).

Studies on the effectiveness of counseling sessions and communication methods are of particular relevance (Michie, et al. 1997). For example, one study found that much of the information regarding diagnoses, risk estimates, and degree of penetrance of genetic disorders provided during counseling sessions is speculative, inconclusive, and marked with a high degree of uncertainty. This was found to conflict with the consumers' desire for certainty (van Zuuren, et al. 1997). Also of interest are evaluations of educational media employed by genetic counselors (Cull, et al. 1998).

The importance of establishing a culturally appropriate toolkit for genetics literacy is supported by the genetics counseling literature, much of which demonstrates a concern for the psychosocial and cultural context within which genetics information and services are provided (De Vos, et al. 1999). Genetics counseling literature also emphasizes the importance of developing a consumer-oriented mode of communication (Smith 1998) that is sensitive to differences in consumer knowledge and attitudes based on socioeconomic status, ethnicity, gender, and education (Fisher 1996; Greb 1998).

Other Health Care Providers

Information on genetics and health care is generated in virtually every medical field, including psychiatry (Baron 1998) and physical therapy (Smith 1999). The current literature review was not designed to systematically review these associated professions. It is clear however that increasing provider awareness of advances in medical genetics is considered to be an urgent challenge in this

literature as well. This theme runs throughout the literature on medical research and provision, demonstrating the influence of human genome research on medical discourse as a whole.

The Public Health Approach

A final subset within the literature on medical research and provision examines the issues of genetics and health care within the framework of public health and public health policy. Public health and public policy approaches have historically encompassed several areas of research that offer complementary perspectives to the primary genomic and medical genetics research generated from the HGP. These areas include but are not limited to: conducting population based research that provides epidemiological data on disorders that have a wide ranging impact on overall levels of public health; developing broad guidelines for screening, testing, diagnostic procedures, and treatment protocols; and, developing policies and initiatives designed to reach out to those sectors of the population that are not currently integrated into the mainstream health care system.

Population-based epidemiological research on genetic-based conditions and disorders provides the data necessary for assessing the collective benefits and risks of new kinds of tests, screening programs, or health education initiatives (Burris, et al. 2000; McNicholl, et al. 2000). The development of tests and treatments for rare genetic disorders will continue at a rapid pace. However, given that these disorders affect relatively small numbers within the general population, their effect on the overall health of the population will be limited (Holtzman and Marteau 2000). It is important to note however, that evaluations of health interventions or screening programs that target for hemochromatosis (Omenn 2000) and relatively rare disorders such as hereditary breast cancer (Lerner 1999; Vineis 1997), and Huntington disease (Holtzman and Shapiro 1998), can provide insight into potential problems associated with screening and other interventions that address more common, often chronic disorders.

Recent primary genomic and medical research has made gains in understanding the hereditary components of more common disorders such as asthma, diabetes, certain forms of cancer, cardiovascular disease, hypertension, and obesity. Given that current research suggests that the degree of penetrance of genotypic risk factors for many common disorders is statistically low, some experts have argued that the new genetic tests for polygenic disorders will have little effect on measurable health indicators in the public (Holtzman and Marteau 2000). This perspective serves to reinforce the traditional public health approach that focuses on environmental and behavioral factors associated with common disorders. Yet, despite the skepticism expressed in some studies, it is possible that the new findings from genomic and medical genetics may provide a complementary perspective (Khoury 2001). The integration of these two approaches has the potential for creating a biomedical approach that is sensitive to the complex

interactions among genes, environment, and behavior throughout the life cycle. And presumably, if studies should ultimately demonstrate scientifically that genetic factors do not explain the prevalence or severity of chronic disorders, they then serve to strengthen claims for environmentally and behaviorally based interventions.

The integrated biomedical approach being developed within the public health field (Austin, et al. 2000; Omenn 2000) offers many advantages for the development of effective population-based outreach for improving genetics literacy relevant to health choices. As subsequent sections will show, one identifiable trend in the literature on primary genomic and medical research, and in the literature on social and policy analysis, treats the individual consumer as the undifferentiated unit of analysis. The public health approach encourages all relevant stakeholders to situate the new research findings within a broader social context. First, it encourages relevant stakeholders to think in terms of populations and groups as well as individuals. Secondly, it reinforces the need for sensible programmatic solutions to incorporate environmental and behavioral aspects into their message content and treatment protocols of health problems.

At first glance it would appear that the population based research and public health outreach efforts that comprise the public health field would naturally complement one another. Population-based primary research into genetic disorders supplies the data necessary for accurately understanding the range of disorders and conditions that affect a population. Deeper consideration of the issue however suggests that population-based studies often involve a number of ethical and moral issues, due to rigid assumptions about individual characteristics based on group data, especially with regard to groups or communities that have faced stigmatization and discrimination in the past. Given that membership in ethnic, racial, linguistic, or other social groups carries markers of discrimination or stigmatization, any research that associates a particular group with a particular disorder affects all members of the group, even if the disorder is relatively rare within the group. Both Tay-Sachs and Sickle Cell Anemia are prime examples of the difficulties posed by this type of research (Wingerson 1998).

The underlying challenge for population-based research results from the use of social or ethnic categories and ascriptions to demarcate a population that presumably shares certain genetic markers. In some cases, these social categories may incorporate a widely diverse set of populations in terms of genotypes and phenotypes. In limited cases however, the use of ethnicity for demarcation has proven to be relatively effective for certain kinds of single gene disorders. In these cases, such as Tay-Sachs disease among Ashkenazi Jews, or recessive ataxia among Acadians, the linking of disease has been relatively non-controversial. Carrier screening for sickle-cell anemia among African Americans

however, offers a cautionary example of the dangers inherent in conducting this type of research without implementing adequate safeguards against discrimination and violations of privacy.

In theory ongoing research that links the genetic components of complex conditions and disorders to ethnic groups in which some individuals have higher rates of risk may lead to interventions that are tailored to specific groups and delivered in culturally appropriate programs. It will be important to distinguish between ethnic populations and ethnic groups or communities. While the former can only be approximated through the use of scientific measures, the latter remain important for purposes of outreach. Ethnic groups and communities provide the social structure through which culturally appropriate research and outreach associated with genetic disorders may be conducted. The aim is not to link genetic disorders to ethnic groups, but rather to educate individuals about potential genetic risks that they may share within groups or communities that have increased risks for certain kinds of disorders due to genotypic penetration into the population.

The literature within the public health approach offers a valuable perspective on how the discoveries of primary genomic research will affect populations and communities within society as a whole. It is particularly important in that it emphasizes environmental and developmental issues related to genetics and health care. At the same time, however, much of this material is directed to health providers and policymakers. Consequently, its contributions to the ongoing social debates about the long-term impact of new findings in genomic research on individual decision making, and on public debates about the impact of biotechnology have been relatively limited.

Social Science and Policy Analysis

This category includes diverse sources of information on the ethical, legal, and social implications of the developments in biotechnology, medical genetics, and associated health issues. These developments are often referred to in this literature as the "new genetics." Information on these topics is produced and disseminated by analysts including, but not limited to: academics, analysts in private not-for-profit think tanks; policy experts; scientists; industry and community leaders; professional associations; and other relevant stakeholders. The Ethical, Legal, and Social Implications (ELSI) programs of the DOE and NIH have contributed a substantial amount of funding for primary research into this dimension of genomic research, but the material extends far beyond those studies. Given the ubiquitous quality of information about genetics, and given the degree to which new developments in genetics and biotechnology affect society as a whole and medicine in particular, it is of no surprise that the literature is quite extensive.

Many of the subjects and topics covered in this section overlap with the educational efforts and discussions directed at health providers, since the ethical dimensions associated with the control of genetic information are of direct consequence for the consumer-provider relationship. Other subjects and topics may overlap with materials from public-sector agencies, given that policy makers often seek advice and counsel from policy experts in the private sector.

The social and policy analysis category can be divided in a number of ways. Juengst makes a distinction between "professional" and "societal" implications of new technologies (Juengst 1995a), where "professional" implications refer primarily to the consumer-provider interaction and "societal" implications refer to broader public health and policy concerns. These distinctions mark different units of study. One set focuses on the individual consumer and decision making about their health, the other unit is the group in relation to the society as a whole. Research into professional implications represents the mainstream of bioethics literature as it has evolved within the institutions of biomedicine. As such, these tend to be closely linked to the decisions that individual consumers and medical providers face in the ongoing provision of medical services. The ethical issues here surround concerns regarding privacy, informed consent, informed choice, and respect for the rights of the individual. The policy implications include concern for regulatory protections for the individual as a consumer of services and protocols for providers of services.

Research into societal implications focuses on issues such as diversity and cultural competencies as they relate to specific communities or subgroups of society, their historical relationships to genetics and biomedicine, and problems of appropriately integrating these groups as stakeholders into new initiatives related to genetics and health care. Research into these areas relate closely to education initiatives, outreach efforts, and policy planning. A second set may be best described as social criticism. This literature examines developments in genomics and their implications for health care and explores the possible social consequences of implementing new technologies on a large scale within a variety of regulatory regimes. These studies are often more speculative, drawing upon current trends in technology and in society as well as past historical experiences in order to create futuristic scenarios where genetics will have had a major impact in the restructuring of medicine and society as a whole.

There is considerable topical overlap between these different approaches. The bioethics approach may focus on the individual ethical dilemmas but often addresses these issues in terms of shared culture as well, such as in respecting religious beliefs of consumers (Cole-Turner and Waters 1996). Given that these analyses and discussions often focus on the intimate relationship between health provider and consumer, this literature often targets medical providers who may be confronted with these ethical dilemmas. Social critiques and policy analyses

also focus on a combination of issues such as cultural diversity and access, all as part of a broader analysis of the social implications of new technologies.

Some of the bioethics literature suggests that ethical issues are shaped by the very nature of current and emerging technologies. Individual consumers now and in the future will have greater access to information about their genetic makeup, as well as genetic information relevant to their potential offspring, their current children, and their aging parents. New technologies introduce new kinds of knowledge and new ethical dilemmas into the consumer's decision making processes. Yet, how consumers interpret information is often difficult to predict. Hence the literature focuses on developing guidelines for communicating information in ways that do not violate the consumer's autonomy. For example, some materials analyze the ambiguities and complexities of the following subjects: "informed consent" (Andrews 1987); non-directive counseling; pre-implantation diagnoses (Botkin 1998; Penticuff 1996); amniocentesis and chorionic villus sampling; and, the psychological implications of genetic testing for certain types of cancer (Cohen 1998; Donovan and Tucker 2000).

Related to these bioethics discussions are analyses of genetic testing for complex and polygenic disorders. In the current health care environment, individuals and the general public are likely to encounter a host of complex disorders. Primary genetics research on cancer, diabetes, high-blood pressure, asthma, Alzheimer, obesity, and cardiovascular disease has been proceeding rapidly. New tests for genetic predispositions to these common complex disorders are being developed daily. However, from the consumer's perspective, the knowledge that these new tests yield is not inherently constructive or useful (Thomas 1999). Not all individuals may want to know about a predisposition to particular disorders (Chronicle of Higher Education 1998; Vineis 1997). Given that the practical benefits of many of these technologies will not be realized until some time in the future, and given that problems of access to health care remain, these types of analyses are able to create future scenarios in which new technologies merge with unresolved problems, creating new kinds of abuses of technologies. For example, new types of genetic testing and genetic information could easily become a basis for denying access to insurance and health care.

More positive scenarios about genetic testing and genetic engineering are likely to emphasize the importance of genetics for the development of somatic and phenotypic therapies. In these scenarios, genomic research focuses on developing new drug, gene, protein, or enzymatic therapies that treat the phenotypic expressions of genetic disorders, thus rendering the more problematic ethical questions less immediate or even redundant.

Several recurring themes can be identified in this literature. These include but are not limited to the following:

- How should practitioners respond to the consumer's right to know or not know information about their own genetic makeup?
- What obligations do individual consumers have to inform family members, given that genetic information is by its very nature shared?
- How is information on genetic disorders or predisposition to genetic disorders communicated from provider to consumer?

Counseling a consumer about health issues is already fraught with fundamental power asymmetries, given that the expert's control of information is usually superior to the consumers. Delivering that information in a non-directive neutral manner, and subsequently counseling the consumer on the best course of treatment without violating the desire for consumer autonomy is a highly delicate process (Lin-Fu and Lloyd-Puryear 2000). Furthermore, while the principle of non-directive counseling remains the goal within the field of genetics counseling, it may conflict with the goals and aims of primary care physicians and other practitioners who focus more exclusively on positive health outcomes (Fineman and Walton 2000).

A subset of this literature integrates the insights of social science research on diversity and access into the more narrowly defined bioethics literature. In this literature, the emphasis is less on new technologies per se, and more on the observation that consumers are not an undifferentiated group that shares the same attitudes and values as the provider. The consumer's socioeconomic status, ethnicity, gender, and religious beliefs may shape her or his decision making processes (Browner, et al. 1999; Rapp 1999). Hence part of this literature may be aptly described as culturally competent bioethics. Another subset expands beyond the individual decision maker to explore how particular communities or ethnic groups may relate to genetics and research related to it (Dukepoo 1998; George 1998; Mittman 1998; Penchaszadeh and Punales-Morejon 1998; Rapp 1999; Rodriguez 1990). Many African Americans for example, collectively remember experiences about sickle-cell carrier screening which may shape their attitudes towards new kinds of genetic tests as a whole (Bowman 2000; Communities of Color Project). These materials place bioethical issues within a broader community outreach and public health framework. Here the focus is on developing culturally competent delivery strategies and programs as well as making sure that new technologies are more widely available.

Another subset of the literature includes social critiques of the Human Genome Project and related biotechnologies as well as more formal analyses of their policy implications at a broader level. Some social critics elevate these

discussions to consider society as a whole (Condit 1999a; Nelkin 1995; Rifkin 1998; Rothman 1998). These examples focus on the current impacts of biotechnology at a variety of levels. These social critiques often depend on a type of "slippery slope" argument or scenarios designed to shock readers into engaging in broader public debate about how society should use these new technologies. Scenarios about designer babies (Lemonick 1999) for example, or the possibility of new eugenics driven by personal choice and preferences, both provoke larger ethical debates about what constitutes a "healthy" or "normal" person in society at large. A primary concern here is that new developments in technology are outpacing our current efforts to resolve basic ethical problems associated with research on genetics and health care. The slippery slope refers to the future potential for selective abortion or "genetic engineering" to expand beyond serious health disorders to selection for traits that bioethicists and the general public see as problematic, including: gender; height and hair color; "intelligence;" or, other features that are part of the normal variations found in human society.

These scenarios gain their power in part from the ways in which emerging technologies are popularized, as discussed below. Often in these popular descriptions, innovations become newsworthy to the extent that they reflect popular health care issues such as obesity, addiction, cancer, and other disorders that affect large sectors of the population. They also tend to become newsworthy when they suggest profound new insights into what are often viewed as intractable social issues such as violence or differences in "intelligence."

On another level, these types of analyses invoke basic dilemmas about our collective ability as a society to control technology. This is strongly argued by Turney in his discussion of the "Frankenstein" metaphor (Turney 1998), or what is often popularly glossed as "playing God" (Peters 1995, 1997). Some discussions focus on these technological developments and primary research in light of current religious value systems (Cole-Turner 1997; Doerflinger 1999). Other literature in this category concentrates on a wider problem of "genetic determinism," or "genetic essentialism." According to these critiques, the use of short phrases such as the obesity gene or "its in the genes," along with discussions about simple genetic explanations of complex behaviors and disorders represent folk models of heredity, not science (Marks 2001). These analyses call for a more careful and systematic use of terminology as well as a more critical assessment of research into issues that are potentially controversial such as behavioral genetics, research on "intelligence," addiction, and mental illness. These become particularly controversial when researchers use genetic explanations to describe populations or ethnic groups. This literature provides an important contextual background for understanding the very broad

implications of genomic research. At the same time, however, it is difficult to assess the impact of these writings on the public consciousness.

Disability rights critiques offer an important perspective on broader social criticism with policy implications (Parens and Asch 1999). These responses question, not necessarily the technologies themselves, but how the promotion and implementation of these new technologies influences cultural notions of what constitutes "normal" or "healthy" persons. The analyses developed in this literature are fertile ground for examining how arguments that utilize discussions of individual rights, autonomy, and personhood intersect with arguments about the virtues of new technologies for improving individual health and the overall levels of health in society. These analyses raise similar issues as those that focus on problems of access for underserved communities, and both approaches serve as an important reminder that new technologies must always be accompanied by serious social debate and policy deliberation.

Research Sponsors

This literature is generated by a wide variety of government-funded efforts to improve genetics literacy and guide the creation of public policy related to genetics and health. Discussion of individual ethical dilemmas, issues of cultural diversity and access, and the wider social impact of genetics and health care, is being driven forward by the efforts of many government agencies, bureaus, and committees. The information they produce comes in many forms, including: peer-reviewed journal articles; conference and workshop proceedings; educational media intended for health care providers, community members, and policy makers; recommendations and guidelines for offering genetics services; fact sheets; and, research results. Much of this information is also available through Internet websites.

The Ethical, Legal, and Social Implications (ELSI) programs of the NIH NHGRI and the DOE Human Genome Program are a major source of funding for investigations into bioethical and policy-related issues. In 1989, an ELSI program was established in association with the Human Genome Project as a working group to support the National Advisory Council for Human Genome Research. It was designed to proactively address the complex issues surrounding the Human Genome Project by funding studies and engaging the public in discussions of social and ethical concerns for the development of policy guidelines. The structure of the ELSI programs has evolved over the years. Presently, both NIH and DOE fund administratively separate ELSI programs. While both programs channel funding for educational efforts and research on issues of confidentiality and privacy related to genetics, NIH and DOE emphasize slightly different areas of research. The ELSI program of NIH addresses four major topical categories:

1. Privacy and fairness in the use and interpretation of genetic information;
2. Clinical integration of new genetic technologies;
3. Issues surrounding genetics research (e.g. informed consent); and,
4. Public and professional education (National Human Genome Research Institute 2000a).

Research funded by the DOE ELSI program has a slightly different focus on complementary issues:

1. Genetics and the workplace;
2. Storage of information and samples;
3. Education; and,
4. Complex or polygenic traits (Clark 2000).

The primary emphasis of NIH ELSI research is on the ethical, social, and legal implications associated with the introduction of new genetic technology into clinical practice, while the DOE ELSI program emphasizes issues surrounding complex or polygenic traits and genetics in the workplace.

Both programs provide and manage grants for education projects, workshops, research consortia, and policy conferences. The NIH NHGRI devotes five percent of its annual budget to its ELSI program, and DOE earmarks three percent of its Human Genome Program budget for its ELSI program. Studies funded by the ELSI programs cover a broad spectrum of social science and policy-related questions, including the concerns of individual consumers, diverse communities, and society as a whole. ELSI-related funds have supported studies designed to explore the risks associated with genetic testing, increase genetics literacy, analyze issues related to privacy and discrimination, and uncover the broader social, ethical, and legal issues emerging from genomic research (Wingerson 1998).

The websites maintained by NIH NHGRI and DOE Human Genome Program include a list of ELSI grants and contracts that have been awarded, as well as a bibliography of resulting publications and products (DOE-ELSI 2001; National Human Genome Research Institute 2000b). The bibliographies demonstrate the extent to which ELSI funding is supporting and shaping social science and policy research on genetics and health care. Since the establishment of ELSI, bioethicists, social scientists, and medical researchers have increasingly focused on genetics and health care topics. This is due, in no small measure, to the funding provided by NIH and DOE. In addition to supporting the ongoing discussions on such issues and the production of information, the ELSI programs also serve as primary channels for the dissemination of information to

professionals and potentially to the public. For example, the DOE ELSI program has funded a number of public education efforts including, *The DNA Files*, a nationally syndicated radio program on genetics, and the PBS television documentary entitled *A Question of Genes*.

Issues related to consumers at the individual level have been investigated in studies such as Gail Geller's "A Model Informed Consent Process for BRCA1 Testing" (Geller, et al. 1998; Geller, et al. 1995; Geller, et al. 1997a). The NIH NHGRI ELSI program has also funded research on issues of access and cultural diversity in conferences such as Edward J. Smith's "Tuskegee Genome Conference" (Smith and Sapp 1997), and in studies such as Carole Browner's "Use of Amniocentesis by Mexicans and Mexican Americans" (Browner and Preloran Forthcoming; Browner and Preloran; Preloran and Browner 1997). Broader, socio-historical analyses of genetics research and popular attitudes have also been covered by NHGRI ELSI funding, including Celeste Condit's "An Empirical Study of Change in Public Genetic Discourse" (Condit 1999a; Condit, et al. 1998) and Dorothy Nelkin's "Human Heredity in American Popular Culture," (Nelkin 1993; Nelkin 1994a; Nelkin 1994b; Nelkin 1995). Through such funded projects, ELSI support has fostered engagement of a variety of professionals and stakeholders with important issues raised by genomics research at all levels.

ELSI-funded research is particularly sensitive to the sociocultural context within which the Human Genome Project is being conducted. As such, it is an important resource for the development of effective outreach to communities to promote genetics literacy. Policy recommendations and education guidelines emerging from ELSI-funded studies demonstrate an awareness of ethical, social, and legal issues as they relate to individual, community, and social concerns. The expansive number of ELSI publications, workshops, and conference proceedings provide many insights that may aid the development of a genetics literacy toolkit. For example, the ELSI programs have funded conferences addressing the impact of reproductive testing on women (Thomson and Rothenberg 1991; Thomson, et al. 1992), workshops on genetic discrimination (NIH-DOE ELSI Working Group and National Action Plan on Breast Cancer 1996), and issues of privacy and confidentiality (Fuller, et al. 1999; NIH-DOE ELSI Working Group and National Action Plan on Breast Cancer 1997).

Of particular interest is the NIH NHGRI ELSI funded "Genome Technology and Reproduction: Values and Public Policy," a project for developing ethically sound policy standards for professionals, institutions, and the public. Many important issues, including privacy, confidentiality, access to services, and cultural diversity are discussed in the proceedings of "Genome Horizons: Public Deliberations and Policy Pathways," a conference devoted to the "Genome Technology and Reproduction" project (Modell and Hartman 1998). This project directly addresses issues of access and the importance of community dialogue. It is the

source of funding for the "Communities of Color and Genetics Policy Project," a community-based initiative designed to incorporate concerns of African American and Hispanic populations into policy recommendations on genetics and health care (Coe 2000). Findings from this project's focus group phase highlight areas of concern among historically underserved populations.

Educational materials produced for health care professionals may also be of importance to developing effective outreach. The DOE Human Genome Program and the NIH NHGRI ELSI programs have funded numerous projects, studies, and workshops for improving professional education on genetics. The NHGRI ELSI Program funded the creation of a continuing education course for primary health care professionals based on insights and perspectives of a working group of genetics counselors (Fine and Koblenz 1994). The NHGRI also funded Virginia E. Lapham's "Human Genome Education Model Program" (HuGEM), a project that produced a series of educational videotapes on genetics and health care for professionals in occupational therapy, physical therapy, and social work (HuGEM Project 1996; Palincsar et al. 1996). Another example of NHGRI ELSI funded educational material emerged from Cardie Texter's "The Human Genome Project: Human and Scientific Dimensions." This project resulted in the creation of instructional videotapes designed for the continuing education of health care providers (MCET 1993a; 1993b; 1993c).

While all of these projects contribute to the widening pool of information on genetics and health care, the information produced through ELSI funding is not disseminated actively through a single source. Results of ELSI-funded studies are typically published in peer-reviewed journals and books. Much of the information being generated with ELSI funds is subsequently communicated within circles of health care providers and academics. In addition, there does not appear to be any systematic effort to translate or package information from ELSI-funded studies specifically for consumers either as targeted efforts to improve genetics literacy or in the form of community outreach programs based on public health approaches. These materials are publicly available through the DOE ELSI Program (www.ornl.gov/hgmis/resource/elsiprog.html) and the NIH NHGRI ELSI (www.nhgri.nih.gov/ELSI/) websites, but they are not generally available through popular media channels.

Community Outreach and Education Efforts

The literature on community outreach and education initiatives describes programs that focus on improving the delivery of genetics services and genetics information to the consumer. Funding for these initiatives comes from a variety of federal and state agencies, as well as from professional associations and not-for-profit organizations. Some of these initiatives focus on developing programs for community outreach and education. Others focus directly on assessing

consumer knowledge and attitudes toward genetics and health care issues. The first two sections are organized according to the sources of funding while the last section summarizes efforts at assessing consumer knowledge.

Genetic Services Branch and Maternal and Child Health Bureau Efforts

Under its mission, the Genetic Services Branch has identified two objectives of particular relevance to the development of the Consumer Network for Genetic Resources and Services Information. These are:

1. To improve the genetics literacy of the public to enhance understanding of the benefits, risks, and limitations of genetic screening and testing, and the implications of genetic information; and,

2. To facilitate the development of well-prepared health care, social service, and public health professionals capable of communicating the benefits, risks, and limitations of genetic screening and testing and accurately interpreting and appropriately utilizing genetic information in clinical and public health practice.

Historically, the GSB has sponsored local projects to develop materials for families to educate about genetics (California Department of Health Services Genetic Disease Branch 1998). Although these materials may be available for the communities in which they were developed, there are generally not easily accessible by a national audience.

Through the GSB, the MCHB has funded several projects and initiatives focused on educating primary care physicians and other providers on the front line of genetics and health care. Examples of these projects include: the Washington State Department of Health sponsored "Genetics and Your Practice" (http://webct.isu.edu/public/GENETICS/); the University of Wisconsin-Madison Departments of Medical Genetics and Pediatrics sponsored "Wisconsin Statewide Genetics Network for Primary Care Providers;" the City of Hope National Medical Center sponsored "Cancer Genetics Education for Primary Care Providers;" and, the Genetic Alliance sponsored "Partnership for Genetic Services in MARHGN." Most recently, the GSB has funded a contract with Society of Teachers of Family Medicine to establish the Genetics in Primary Care Project (http://bhpr.hrsa.gov/dm/genpc.html). This project is co-funded by the Bureau of Health Professions, HRSA, NIH, and AHRQ, and exists in twenty medical schools across the country. Some projects have developed websites that report their findings. Given that these projects target primary care providers, the materials developed have not been widely disseminated for public consumption.

In addition to these primary care provider education programs, MCHB has sponsored several conferences and workshops. One workshop, held in December, 1999 brought together state legislators from Arizona, Iowa, Kansas, Minnesota, New Hampshire, North Dakota, Oklahoma, Vermont, and Wisconsin to provide them with timely information on topics such as: state legislators' role in genetics policy: genetic privacy and discrimination issues related to children; state newborn screening system design; and, access to needed services for children with genetic disorders. A second workshop entitled "Impact of Genetic Issues on Child Health Policy: A Workshop for State Executive Branch Officials," was held June 21-23, 2000 at the Rensselaerville Institute. The goal of the workshop was to inform officials about the implications of the new developments in genomic research for newborn screening programs that are already well established as part of many state public health programs. This workshop covered a wide range of topics including: general background information on the Human Genome Project; public health approaches to genetics issues; and, a discussion of state policies, roles, and responsibilities. In addition to these general discussions, the workshop provided a number of more detailed presentations of lessons learned from past experiences, including discussions on sickle-cell screening and discrimination, and a case study of how the Massachusetts Department of Public Health Newborn Screening program responded to new tests and testing technology. In addition to the presentations, the workshop provided extensive supporting and background materials for participants. Materials from these conferences have not been widely disseminated for public consumption.

Centers for Disease Control and Prevention Efforts

The CDC has also developed substantial materials linking genetics to public health efforts. The genetics section of the CDC website (www.cdc.gov/genetics) offers overview information on scientific and technological developments, as well as "consumer friendly" sections such as "Frequently Asked Questions about Genetics and Public Health" (www.cdc.gov/genetics/publications/faq.htm). The CDC Office of Genetics and Disease Prevention provides a set of resources on its website (www.cdc.gov/genetics/resources/grevolution.html) entitled "A Public Health Perspective." This site provides reprinted scientific articles, editorials, and news articles relevant to the current issues. The most current listings include discussions of "Will Genetics Revolutionize Medicine?" (Holtzman and Marteau 2000). The site offers a convenient way to trace this particular debate about genetics and health care as it has developed in the literature.

The CDC hosted a conference entitled "Genetic Competencies for the Public Health Workforce," held August 16-17, 2000. In addition to providing background information on public health workforce development, the conference materials include descriptions of competencies in essential service areas, and for

individual members of the public health workforce (Centers for Disease Control and Prevention 2000).

The CDC and the public health approach place greater emphasis on the implementation and practical application of new genetic technologies on a broader scale than just individual treatment. In addition, the emphasis on community health and outreach, as suggested by the competency guidelines and the other materials published on the CDC website, provide fertile ground for research design in support of the development of effective population-based outreach.

Efforts of Other Associations

Beyond the federal agencies and programs are a variety of state and regional genetics associations that inform policy development by contributing information on genetics and health care. These networks are funded in a variety of ways. The Texas Genetics Network, for example, is funded by MCHB and several state public health departments in south central United States. Similarly, the National Newborn Screening and Genetics Resource Center (NNSGRC) created through a cooperative agreement between MCHB GSB, HRSA, and the University of Texas Health Science Center at San Antonio, serves as a national resource center for information and education in the areas of newborn screening and genetics (http://genes-r-us.uthscsa.edu/). Some, like the Mountain States Regional Genetic Services Network, are private not-for-profit entities, while others, including the Mid-Atlantic Regional Human Genetics Network and Pacific Northwest Regional Genetics Group, are affiliated with academic institutions. A number of these networks are supported directly by the Genetics Services Branch. These networks function to develop and promote guidelines and standards for the provision of genetics and health care information and services. They also create and disseminate educational materials for health care professionals designed to increase genetics literacy and provider skills in handling the growing demand for genetics based health care services (Pacific Northwest Regional Genetics Group 1995). Information disseminated by these networks may be useful for community outreach and education efforts.

Numerous professional associations also establish guidelines and standards for the provision of genetics services and information. A number of these associations deal specifically with genetics and health care. The American Society of Human Genetics, for example, establishes guidelines for the development of medical school curricula in genetics (American Society of Human Genetics Information and Education Committee 1995) and issues statements on new developments in gene therapy (American Society of Human Genetics Board of Directors 2000). The American Board of Genetic Counselors provides certification for genetic counselors and issues standards and requirements for graduate programs seeking accreditation for genetic counseling programs

(American Board of Genetic Counseling 1996; Fiddler, et al. 1996; Katsichti, et al. 1999).

Efforts to incorporate genetics-based health education into medical school curricula and to guide genetics training are also pursued by professional organizations such as the American Academy of Family Physicians (American Academy of Family Physicians 2000), American Society of Clinical Oncology (American Society of Clinical Oncology 1997a; 1997b), and the International Society of Nurses in Genetics (Anderson, et al. 2000). The American Academy of Pediatrics is active in developing newborn screening protocols (1992; 1994b; 1993) and issuing guidelines for the prevention and treatment of genetic disorders (1993; 1996c; 1999). Numerous other professional associations engage in efforts to inform primary health care providers with accurate, up to date standards for the prevention, diagnosis, and treatment of genetic disorders. These include the American College of Medical Genetics, the American College of Obstetricians and Gynecologists, the American College of Physicians, the American Medical Association, and the American Public Health Association.

A number of professional associations have devoted significant resources to promote genetics and health care by reaching across disciplinary boundaries. There are two initiatives in particular that attempt to integrate all of those involved in genetics and health care under broader umbrella organizations. The National Coalition for Health Professional Education in Genetics (NCHPEG) has as its goal to "achieve consensus among health professional leaders regarding the core genetics competencies that all health professionals must have and stimulate their adaptation by different health professional disciplines" (National Coalition for Health Professionals Education in Genetics 2000). The Core Competency and Curriculum Working Group of NCHPEG developed a comprehensive set of competencies in the knowledge, skills, and attitudes areas of genetics education, and disseminated these guidelines through a variety of media, including formal publications, websites (www.nchpeg.org), and press releases.

The second initiative, Genetic Resources on the Web (GROW) represents a wide variety of public and private sector groups including but not limited to: government organizations such as CDC; MCHB; NIH NHGRI; DOE Human Genome Program; professional associations such as AMA, ASHG, ABGC; and, private sector groups such as Celera Genomics, Pharmaceutical Research & Manufacturers of America, and DNA Dynamics, Inc. The participants in GROW all share a common objective to effectively use the Internet to disseminate high-quality information about biotechnology, genomic research, and genetics and health care. GROW is still in the early stages of development. The group is currently working towards resolving funding issues, developing guidelines and standards to ensure high-quality information, and creating a comprehensive search engine that would link all participating sites. If completed, the GROW

search engine would be a valuable resource for the dissemination of a wide variety of information on genetics and health that would clearly support the development of effective outreach.

The various projects and initiatives described above all represent efforts on the part of organizations from various sectors of the health care field to introduce regulatory mechanisms and guidelines necessary for effectively implementing the new technologies and innovations resulting from the Human Genome Project. A comprehensive summary and discussion of proposed nation-wide policies and guidelines can be found in the report: "Enhancing the Oversight of Genetic Tests," by the Secretary's Advisory Committee on Genetic Testing. The report is a result of coordinated efforts on the part of a wide range of participants to create oversight mechanisms for new genetic testing. The report covers issues related to testing, including: efforts to determine the risks and benefits of particular genetic tests; ensuring that individuals and family members have "access to appropriate genetic education;" ensuring that principles of informed consent are followed; creating mechanisms for monitoring clinical standards; and, suggesting legislation designed to "prohibit discrimination in employment and health insurance based on genetic information" (Secretary's Advisory Committee on Genetic Testing 2000).

While the bulk of the initiatives and projects within the social science and policy analysis literature generate educational materials, practice guidelines, and policy recommendations for providers and other stakeholders, a smaller subset of materials explores the knowledge, attitudes, and beliefs of consumers. The following section offers several examples of this research.

Consumer Research

Research on consumer knowledge of and attitudes toward genetics and health care provides baseline data for assessing consumer needs and concerns. Some of these studies are designed to equip health care providers with an understanding of the challenges associated with communicating information on genetics (Andersen, et al. 1997; Ludman, et al. 1999; Schover, et al. 1998). Other studies offer suggestions for outreach and policy development based on a heightened awareness of consumer needs and concerns (March of Dimes 1992; Moyer, et al. 1999; Ose, et al. 1998). The findings within both categories constitute an important resource that may aid the development of a genetics literacy toolkit. The following examples illustrate that consumer perceptions influence how information on genetics and health is incorporated into health care decisions.

Richards and Ponder (1996) demonstrate that folk definitions of inheritance, based on notions of kinship and descent, are at odds with scientific explanations of genetic inheritance. According to this study, genetic relatedness is conceived

in accordance with the strength of kinship ties and obligations between categories of kin. For example, subjects in the study underestimated their genetic relatedness to those with whom they have weaker social kinship ties, such as sisters, uncles, and grandmothers. This study suggests that folk ideologies of inheritance may impede the understanding of scientific findings, and may complicate the communication and calculation of genetic risk factors in medical decision making. Jonathan Marks discusses four categories of folk ideologies of heredity in detail, illuminating the misconceptions associated with such constructs. He warns, however, that these folk ideologies do not belong exclusively to the "uneducated" public. According to Marks, many scientific studies on genetics and behavior are based on the same assumptions, suggesting that, "geneticists have assimilated the same folk ideas as everyone else" (Marks 2001:64) Implications of these findings include the need to know such biases prior to developing educational protocols, and the need to consider bottom-up and community- based approaches to genetics education.

Studies designed to assess consumer interest in genetic testing may also attempt to elicit the correlation between attitudes and knowledge. One study based on a statewide telephone survey of Kentucky residents demonstrated that a lack of interest in genetic susceptibility testing is associated with less education (Andrykowski, et al. 1996). This does not mean, however, that increasing the genetics literacy of consumers will necessarily encourage greater acceptance of genetic testing. Results of a study in Finland on the relationship between consumer knowledge of and attitudes towards genetic testing provide lessons for guiding genetics literacy initiatives (Jallinoja and Aro 2000). Subjects in the study with the lowest level of knowledge demonstrated the greatest difficulty taking a position on attitude statements. However, a higher level of knowledge and understanding of genetics did not predict unequivocal support and enthusiasm for genetic testing. In fact, one of the significant findings for the development of outreach is that greater genetics literacy, though associated with a higher level of acceptance, is also associated with greater levels of suspicion and uncertainty.

In fact, the Survey Research Center at the University of Maryland conducted a survey of peoples' attitudes before and after participation in the conference "The Human Genome Project: Reaching Minority Communities in Maryland." Although participants perceived that they learned more by participating in the conference, the overall percentage of respondents who believed that the benefits of genetics research applications are greater than the harmful results for members of ethnic minorities decreased from 65.7 percent to 53.1 percent (Survey Research Center 2001:115). While the conference raised awareness and knowledge of the role of genetics in health care, it also raised doubts that genetics services will benefit historically underserved populations.

More broadly, these findings question the assumption often expressed by experts in the field of biomedicine that a knowledgeable public will naturally promote advances in genetics health technology. This assumption is challenging to the development of effective community-based, population-specific outreach. It promotes a top-down model of education that treats people's preexisting knowledge, beliefs, and attitudes as potential flaws in need of correction, and it systematically disregards the effect of peoples' knowledge, beliefs, and attitudes on decision making about health concerns. (Kerr, et al. 1997; Rothman 1998).

There is significant interest among consumers for information on genetic testing and its implications. The Kentucky survey found that 87 percent of respondents expressed a high level of interest in learning their personal genetic predisposition for cancer (Andrykowski, et al. 1996). This finding is supported by results of a nation-wide telephone survey conducted by Harris and Associates for the March of Dimes Birth Defects Foundation (March of Dimes Birth Defects Foundation 1992). The survey found that 72 percent of the 1000 respondents would take genetic tests to determine whether they or their children would be susceptible to serious or fatal genetic disorders. These studies, though currently almost five to ten years old, are important for understanding the degree of public support for genetics and health care research and for illuminating general public attitudes and knowledge. More recent public opinion surveys that provide data on attitudes towards genetics and health information sources can be accessed through the web-based Lexis-Nexis Academic Universe search engine (www.lexis-nexis.com).

The use of telephone interviews to assess public perceptions, however, may not provide an understanding of the divergence between stated level of interest and actual performance. For example, a study reported by Lois Wingerson (1998:130) explored attitudes as well as decisions regarding testing for BRCA1 among members of families at risk for developing breast and ovarian cancer. In the study, tested subjects participated in educational sessions and were given the opportunity to learn the results of their screening. Forty percent of the subjects did not complete the education sessions, and 60 percent declined to learn the results of their screening. Follow-up questions revealed that subjects were most concerned about the accuracy of the tests and the possibility of losing health insurance coverage if a positive test result was reported.

Understanding public concerns and attitudes is a cornerstone of a number of initiatives designed to offer policy recommendations and increase genetics literacy. The Communities of Color and Genetics Policy Project, funded by the ELSI Program of the NIH NHGRI, is being conducted by the University of Michigan, Michigan State University, and Tuskegee University. This project began with a focus group phase designed to elicit statements from members of African American and Hispanic communities regarding genetics research. Issues

of concern were discussed before and after an educational piece provided by the facilitator. The findings of these focus groups provide insights into the needs and concerns of members of these particular minority communities. The report highlights a number of these concerns including: the desire to have a voice in the policy making process; general distrust of the government; ethical pitfalls of cloning technology; insurance and employment discrimination based on genetic test results; and, the importance of privacy. The report notes differences between the African American and Hispanic focus groups, such as the greater degree of government distrust among African Americans.

A more focused study compared the difference in knowledge of breast cancer and cancer genetics among African American and Caucasian women awaiting routine medical service (Donovan and Tucker 2000). A significant finding was that African American and Caucasian women perceived the risks and benefits of genetic testing differently, and that this difference was based on the unique psychological, social, and economic concerns of each group. Similar studies have been conducted on perceptions among Ashkenazi Jewish breast cancer patients (Phillips, et al. 2000), and on Southeast Asian attitudes toward screening and prenatal diagnosis (Yuen, et al. 1988). The National Dialogue on Genetics project offers other diverse ethnic perspectives (Mittman 1998b). These reports can serve to enhance awareness of cultural diversity and its influence on attitudes and behaviors, and are guideposts for the research designed to reach underserved populations.

Balch Associates conducted another important focus group study on perceptions of genetics research for the National Health Council as part of a genetics literacy project (1999). The study supports the notion that there exists a "buzz" of information in the public domain on genetics research, but that people do not have a specific bounded conceptual category for "genetics research." Participants did not demonstrate a comprehensive view of the information on genetics received through mass media, and remembered only fragments of the information received in the past. Knowledge of genetics was often inaccurate. Participants were most concerned with personal issues of privacy and confidentiality and the broader, social and ethical implication of genetics research, such as the moral dilemmas associated with human cloning. The specific findings of the study offer information on a wealth of topics: what people want to know about genetics; attitudes toward genetic testing; and, channels through which information is transmitted. An interesting finding is that healthy participants were not actively seeking information on genetics research, but were passively receiving it through mass media such as television, newspapers, and the Internet. People who either had a genetic disorder, or a family member with one, were more likely to seek information on genetics and health care by subscribing to health organization newsletters, visiting medical centers, and exploring the Internet. This is not surprising. As described below, people

meaningfully engage information on genetics and health when such information becomes personally relevant.

Overall, the findings of studies on consumer perceptions underscore the importance of understanding the broader sociocultural framework within which genetics and health care information and services are offered. They also highlight the need to be aware of the attitudes, knowledge, beliefs, and concerns of consumers. Although many of such studies advocate efforts to increase public and consumer knowledge of genetics, this review suggests that such efforts targeted specifically to increased knowledge in a biomedical sense may not result in a heightened consumer interest and confidence in genetics services. These studies provide ample evidence that education is necessary for encouraging informed, autonomous decision making among consumers. However, they also suggest that attempts to educate people based on the mission to make genetics services "acceptable" run the risk of eroding the autonomy of consumers.

Journalism

Journalism constitutes the final category of information covering the broad implications of new advances in biotechnology and genomic research. The literature review culled information from many popular news media channels including newspapers, magazines, books, and websites. This information covers an expansive array of topics, reaches a large and diverse audience, and contributes to the overall societal buzz surrounding genetics and health care. Sources of journalistic information on genetics include printed mass media, e.g., *Time*, *Newsweek*, daily newspapers, health magazines, books, radio, television and cable news channels, news wires and their Internet sites, e.g., Reuter's Health, as well as niche media that target health professionals and interested consumers, e.g., HMS Beagle (www.biomednet.com/hmsbeagle/), Geneletter (www.geneletter.com). An analysis of journalistic media is essential to understanding the challenges of producing information for large audiences.

New research findings in the field of genetics and health are constantly being channeled through the popular media. These reports often include discussions of the ethical, social, legal, and financial implications of these findings. Information on genetics in journalistic media reports covers many topics. For example, one issue of *Time,* a popular news magazine, featured articles on "The Biotech Century" that covered: the history of the Human Genome Project; prenatal genetic testing; genetic "fingerprinting;" eugenics; gene therapy; cloning; genetic profiling; and, genetically modified foods (January 11, 1999). In addition, consumers find information about genetics and health issues in a variety of reporting categories, such as: business and biotechnology; sports; health and fitness; science and the environment; and, religion and ethics.

According to a focus group study conducted for the National Health Council, journalistic mass media is the primary mode through which members of the general public receive information on genetics and health care (Balch Associates 1999). However, there are numerous challenges for effective genetics education associated with how information is disseminated through this channel of communication. The lack of consistent reporting on any one topic may prevent consumers of mass media from forming a systematic and comprehensive understanding of genetics and health care. In fact, the NHC focus group study found that people do not have a specific cognitive category for "genetics research." This may result, in part, from the inaccuracies in reporting, and the often fragmented and inconsistent nature of information communicated through mass media channels by science writers.

Journalistic media faces the challenge of distilling and translating highly technical and complex research findings for mass consumption. Science writers often report on a diversity of topics with which they are not always familiar. (McGowan 2001). Inaccuracies in reporting are a significant source of confusion for recipients of this information, and may contribute to gross misunderstandings. Terms such as risk have very particular meanings in science and biomedicine. Science writers may adopt such terms without attention to the subtleties of specialized usage. For instance, the statistical basis of risk assessment is not carefully communicated in science writing for popular consumption when reporting on "risks" for conditions and disorders associated with genetics. Headlines that herald the discovery of "genes for" cancer may hinder consumer understanding of genetic processes. These representations of cancer may support biologically inaccurate notions of heritability, thereby creating the false impression that cancer is simply passed from one generation to the next, and that an individual is doomed by virtue of "having" the "cancer gene" (Rothman 1998). This example also highlights the potential dangers of genetic essentialism, a common theme in popular representations of gene-linked disorders (Nelkin 1995).

Differences in reporting research findings across channels is another source of confusion for consumers. The same research findings may be reported by the media in a manner which could lead to diametrically opposed interpretations in by consumers. For example, the headline of a *Washington Post* article on a Scandinavian cancer study announced "Cancer Study Deemphasizes Genes' Role" (July 13, 2000:A1). The headline of an Associated Press article covering the same research that ran in the *Baltimore Sun* on the same day, however, proclaimed, "Genetic Factor in Cancers Strong, Researchers Find" (July 13, 2000: 6A). Both articles employed the same statistics and quotations to support opposing claims. Inconsistent reporting such as this may contribute to misunderstandings or misinterpretations. Some readers may be confused by multiple interpretations of the same research findings. Other may develop well

structured yet scientifically or medically inaccurate perceptions which could influence their decision making processes about health and their health care choices.

Overcoming inaccuracy and inconsistency are not the only challenges to science journalism. Market forces are one of many determinants of what is considered newsworthy by the media, and they affect the type of information disseminated and the tenor of its reporting. Biased coverage of issues in the media may cloud public understanding of genetics and health care by focusing on high profile disorders and controversial theories about the genetic determinants of human behavior, while offering scant information on less marketable but no less important topics. For example, cancer coverage in women's magazines favors reporting on breast and skin cancer, and rarely offers information on the diagnosis, risks, and prevention of lung and colon cancer (Gerlach, et al. 1997). This suggests that attempts to improve genetics literacy should be based on an awareness of the information consumers are receiving, as well as the information they are not receiving.

Content is only one dimension of what consumers are and are not receiving from the producers of mass media. The language used to describe genetic research and its implications for consumers is of equal importance to understanding how consumers may be misinformed about genetics and health. Some social scientists emphasize that popular reports on advances in genetic technology employ language that tends to reduce the complexity of life to a genetic essence (Nelkin 1995). In this view, the use of the 'blueprint' metaphor to describe the human genome may be interpreted by individuals as reducing identity, behavior, and future health possibilities to an unalterable genetic script. In the most extreme scenarios depicted by critics of genetic essentialism, genes become the excuse for social ills including poverty and crime, the potentially misleading scapegoat for parents of children with behavioral problems, and the justification for inequality and maintenance of the social hierarchy (Nelkin 1995).

Others, however, argue that the high degree of ambiguity in reports on genetics and health care may result in greater awareness of the interaction between genes and the environment, thereby diminishing the negative impact of genetic essentialism on public opinion. Condit, for example, provides a sophisticated, historical analysis of the treatment of "genes" in the popular media (Condit 1999a). Media sampled between 1980 and 1995 demonstrate an overall reduction in the degree of determinism assigned to genes, and a greater emphasis on: the genetic "contribution" to disease; the role of behavior in increasing personal risk above a normative mean; and, the ability to manipulate the genetic makeup through scientific advances. A survey by the same author designed to elicit public perceptions of the role of genes in determining behavior

revealed that the blueprint metaphor is interpreted as a malleable, probabilistic forecast, not an unavoidable fate (Condit 1999b).

While the effect of these representations on public perception is debatable, social scientists tend to agree that the use of sensationalist and reductionist language in the popular media is problematic. The genome has been portrayed as the "Holy Grail" of biology's quest to unravel the "secret of life," as the "Bible" and the "Book of Man" (Nelkin 1995 p.39). Advances in genetic testing and diagnosis have been likened to "playing God" (Peters 1995), and a great deal of press has emphasized the limitless potential of genetic technology for improving health (Micklos and Carlson 2000). Although this language is transmitted to people via popular media, scientists often produce it, though with unintended consequences. In a 1989 interview with *Time*, James Watson proclaimed that, "We used to think that our fate was in the stars. Now we know, in large measure, our fate is in our genes" (Jaroff 1989). Messages like this may be intended to promote the Human Genome Project, but they also contribute to public misunderstanding about genetics and health. According to the science editor of the New York Times, the burden of responsible reporting partially falls on the shoulders of research scientists themselves (McGowan 2001). This constitutes a valuable lesson for developing effective outreach. As participants in the creation of information on genetics and health care, we must be aware of the power of language to influence public perception, and assume the burden of responsible reporting.

Analysis of popular media, in addition to illustrating the importance of carefully crafted language, also provides lessons on the form and narrative structure of genetics and health care information. According to Condit, popular media typically portray new genetics services as products in the consumer domain, creating an emphasis on personal health and fitness (Condit 1999a). Information presented in personally relevant formats to individual consumers may serve to heighten public awareness of genetics issues. Although we do not know the effect of consumer-oriented journalism on individual knowledge and decision making, it does have the potential to enhance autonomy by increasing genetics literacy relevant to decisions about one's health.

A significant challenge presented by the dissemination of knowledge for mass consumption such as consumer guides for improving personal genetic health, is the difficulty in capturing the needs and concerns of socioculturally diverse populations. This form of journalistic information often targets an undifferentiated male or female consumer, thereby ignoring those topics that concern members of historically underserved populations. Public health initiatives that focus on community outreach address the need to understand cultural diversity for making genetics and health care personally relevant. The design of a toolkit for genetics literacy must take these lessons into account

substituting culturally specific and appropriate approaches for the production of information designed for mass consumption.

There are sources of information that yield quality information on genetics and health care that may be useful resources for developing effective outreach. For example, GeneTests (www.genetests.org) is a not-for-profit organization funded by the National Library of Medicine and MCHB that provides accurate information and valuable resources on genetic testing and counseling. A number of websites sponsored by for-profit companies, including GeneSage's geneletter.com and BioMedNet's online magazine HMS Beagle, also publish information on genetics and health care. These sites tend to provide balanced, comprehensive, and objective reports on technical research as well as the ethical, social, and legal issues at stake. While these channels provide a high-quality of information that is likely to increase genetics literacy, they reach audiences actively seeking information about health but do not directly reach the general public.

The review of information produced in the sphere of journalism provides a number of lessons to guide the development of effective outreach. While science journalism tends to reach a broader audience, translating complicated research findings into information that is relevant to, and understood by, diverse consumers creates barriers to improved genetics literacy. Making information accessible requires using accurate and responsible language that does not oversimplify genetic processes. Making information interesting does not always generate fair, balanced coverage of issues. While the mass media's emphasis on personal health genetics may encourage engagement with genetics issues and promote autonomous decision making, science journalism tends to neglect the sociocultural context within which health care decisions are made. The format of popular media reporting tends to treat the audience as undifferentiated, individual consumers, glossing over the many issues of relevance to underserved populations.

Conclusion: A Brief Analysis of the Categories.

Information about genetics is ubiquitous. Consequently it is no surprise that the five categories used above overlap thematically. In addition, producers may disseminate information that is relevant for several of the above categories. For example, developing new standards of clinical care for amniocentesis incorporates elements of genomic research, the provision of medical services, and the formulation of government policy. Ethical concerns associated with decisions about pregnancy, and issues related to raising a child with a heritable disorder must also be taken into account. While closely linked, the categories used here capture major differences that will be important for developing effective outreach.

The first two categories of information production and dissemination, "Primary Genomic Research and Biotechnology" and "Medical Provision and Treatment," overlap in reporting research findings on technical innovations that relate to primary research. The primary research for medical providers overlaps with basic human genome research, but it is also more specific, focusing almost exclusively on the discovery and mapping of particular loci that are associated with single gene disorders and the multiple loci associated with complex or chronic disorders.

The potential that genetics has to radically change medicine is clearly recognized in the literature. The development of new tests for single gene disorders, historically referred to as Mendelian genetic disorders, and the ongoing identification of candidate genes involved in common or late onset disorders are just two factors that should move medical providers and consumers to increase their awareness and to improve their genetics literacy. In addition, new technical innovations driven by biotechnology have in turn prompted serious discussions about the ethical, social, and legal implications of genomic research (Rifkin 1998; Wingerson 1999). These developments do not occur in isolation, although they are often produced and disseminated in the private sector. As a result, economic forces are at play, and attention is drawn in new ways to the ethical issues, particularly as these developments are communicated to the public. Through federal funding, the Ethical, Legal, and Social Implications programs of the DOE Human Genome Program and the NIH NHGRI are promoting discussions among stakeholders and major actors to address the interplay of private and public concerns.

The medical provider literature also includes two other subsets of information production and dissemination. One addresses what some experts in the field see as an impending gap between the genetics competency levels of medical providers and the latest findings and innovations generated by the Human Genome Project. Much of this material embodies proactive efforts on the part of professional associations and other organizations to educate specialists, primary care physicians, and other medical providers other than genetics specialists, about the ongoing innovations in testing, diagnosis, treatment, and care of conditions and disorders that have a genetic component. The second subset, generated from the field of public health, provides a valuable complement to the disease-based research that is oriented toward individual treatment. The public health literature has consistently sought to link primary genomic and medical research to epidemiological and population-based research into genetic disorders. The traditional public health focus on environment and behavior has stimulated discussions about how genetic and environmental interactions affect phenotypic expressions of genetic disorders. In addition, the public health field has historically addressed issues of access for all members of society, including

underserved communities. Their work in this area offers an important bridge to social and policy analyses that are concerned with these same issues.

The next two categories, Social Science and Policy Analysis and Community Outreach and Education Initiatives are broadly linked by a commitment to make the genetics part of sound health policy and practice. It is recognized however, that defining "sound policy" is challenging given the cultural, religious, and ethnic diversity of the United States. Nevertheless, efforts are clearly targeted toward finding a balance among multiple views and value systems, and bringing all relevant parties into a meaningful discussion on policy and practice.

Social and policy analysts address the ethical, social, and legal implications at a variety of levels. Social and policy analysts include bioethicists, social scientists, policy makers, politicians, and scientists who are acting as public representatives of their field. The degree to which their analyses comments or makes policy recommendations on the ethical, social, and legal implications of new scientific and biomedical developments varies. Many analysts address concerns that individuals and couples must face, such as:

- Should a pregnant woman have amniocentesis testing (Gekas, et al. 1999);
- What should one do if the test results are positive for a particular disorder or condition (Bourguignon, et al. 1999);
- Given prior knowledge of a genetic disorder, should an individual choose to have children naturally, should they rely on pre-implantation diagnoses and *in vitro* fertilization, or should they forego childbearing entirely (Schover, et al. 1998); and,
- Will their insurance cover the test or the procedure? What will the insurance company do with the information if they gain access to it (Billings 2000b)?

Broader social questions have also received substantial attention. These include the ownership of the genome and patent law (Eisenberg 2000); the use of new genetic technologies for "enhancement," or "positive eugenics" (Richter and Bacchetta 1998); or, whether or not insurance companies should deny coverage to individuals with "genetic predispositions" (Ad Hoc Committee on Genetic Testing Insurance Issues 1995)

Social scientists and policy analysts consider how research in the scientific and biotechnology sectors effect change in other sectors of society. For instance, how might new developments in these sectors conflict with or contribute to areas of ethical conflict such as abortion, eugenics, or discrimination based on a variety of factors. In this literature, researchers try to understand and anticipate the

effects that new scientific, technological, and medical developments have on consumers.

Social scientists and policy analysts engage these questions from three perspectives. The first considers the difficulties of balancing principles of individual autonomy and rights to privacy with those of medical necessity or the larger public good. For example, they may ask questions regarding the kinds of counseling a carrier with a genetic disorder should receive regarding reproduction, or they may discuss the strengths and limitations of the idea of "informed consent" regarding genetic testing.

The second type of inquiry focuses on the impact of new developments on society more generally. Examples of this basic approach include, but are not limited to, questions regarding: intellectual property issues; developing federal guidelines for the screening and testing of chromosomal and genetic disorders; developing guidelines for stem cell research; conducting studies among ethnic groups regarding their attitudes toward genetics and medicine; and/or, determining who should have access to information about an individual's genetic makeup.

The third perspective within social and policy analysis overlaps with public health concerns. Here questions are raised about issues of cultural diversity, problems of access, and the historical experiences certain ethnic groups or other underserved communities have had with biomedicine and genetics based health care. This approach overlaps and contributes to the more general social analyses regarding eugenics and the implications of genetic engineering, but it also provides valuable focus on individuals as engaging with medical and ethical concerns through a highly diverse set of knowledge, attitudes, and values.

The producers of information in the first four general categories disseminate information in a wide variety of media. Some forms are targeted to specific audiences, while others appeal to broader audiences. For example, technical information and primary research findings are disseminated in highly structured and controlled formats such as professional peer reviewed journals. Virtually all of these groups have also been aggressive in their use of the Internet, as complex and fragmented as it may be, and all of them indirectly use the popular media through newspapers, popular magazines, radio, and television.

Hence, in addition to these four categories of professional experts there is a final group that deserves special attention. Science writers and journalists are a primary means of production and dissemination of information about genetics and health to the public. The category of science, health, and biotechnology writers take the rather specialized information generated in molecular biology,

medicine, and biotechnology research and "re-package" it so that non-expert consumers understand its relevance.

The dividing line between this type of science journalism and other categories of information production and dissemination is often fuzzy. The distinction is one of form and placement. Articles tend to be shorter, found in more popular media outlets, and tend to focus on newsworthy events such as technological innovations, or new "hot button" research findings. This genre is consumer driven, and presumably is more responsive to the desires and concerns within popular culture at any given moment. It may even heighten consumer desire for information to address concerns about genetics and health through emphasizing highly marketable stories. More than any of the other categories, this genre of information contributes to the "buzz" about genomic research and biotechnology. As such, the various communities of experts who are more intimately engaged in research, medicine, and policy analysis have an ambivalent relationship with this category of information production and dissemination. While they are often dissatisfied with the particular topics or modes of translation that journalists use, they must also rely upon science writers to publicize their work. The translation may not be effective, but this feedback loop, from research to popular media, is the primary means whereby information about genetics and health reach the public.

Scientific journalism translates information about primary research findings into language deemed acceptable and accessible to the public. Much of the information produced and transmitted in the previous four categories is of a highly specialized nature. Nevertheless, given the degree to which such research may affect society on a broader level, or affect individuals and families at the most intimate level, these findings are by their very nature, newsworthy and important for dissemination to the general public.

This highlights several essential questions that this project seeks to address: what does the general public know and understand about genetics; through what channels do they obtain information; what is the minimum level of knowledge that the general public requires in order to make informed decisions about their health as new developments in genomic research and biotechnology change the biomedical view of health and the provision of medical care; and, how can this information be transmitted most effectively?

Meaningful Engagement with Information on Genetics and Health

Question 2: When do people encounter information about genetics and health care?

Findings of the literature and materials review discussed in the previous section indicate that information on genetics and health care is ubiquitous. It is generated by multiple sources and disseminated in a variety of forms to diverse audiences. These findings will aid the project partners by providing an understanding of the kinds of information available and the challenges presented by their production, form, and content. This research is also guided by the need to understand when or under what life circumstances people encounter this information on genetics and health care. Awareness of the factors that contribute to engagement with this information will provide guidance for the development and application of the genetics literacy toolkit by highlighting the life events and processes that encourage people to learn about the relationship between genetics and health.

This analysis will demonstrate that, while people encounter information about genetics and health care on a daily basis, the degree to which they engage this information varies in accordance with a host of personal life events, circumstances, and external forces. There is a continuum of engagement, ranging from apathy to activism that varies among individuals and communities and across the life cycle. Understanding the forces that shape the active engagement with genetics and health care information may suggest future means by which the genetics literacy toolkit can be meaningfully implemented to promote informed, autonomous health care decision making.

Information generated by popular media representations and reports constitutes the buzz on genetics and health care that is encountered on a daily basis. People may encounter this background of information unintentionally through many channels, including newspapers and magazines, news broadcasts, and everyday conversation. These encounters are often inconsistent, contributing to a fragmented, incomplete knowledge of the relationship between genetics and health. The encounters may focus attention on the sensational, controversial aspects of genetics research and its future potential, rather than provide knowledge of the current, practical integration of genetics and health care. Discourse on genetically modified foods, cloning, stem cell research, and other morally charged issues contributes substantially to the buzz of information.

While this discourse may encourage greater engagement, the relationship between genetics and health care may be lost in these reports. Because such encounters are not systematic, they may prevent people from perceiving genetics research as a separate category of science with health care implications (see Balch Associates 1999). Thus, while the buzz may increase awareness of genetics, attending to it does not necessarily encourage individuals to seek further information on the implications of new research in genetics for their personal health. Furthermore, attending to the buzz does not necessarily empower individuals with the knowledge they need to make informed health care decisions. This provides a lesson for research: information on genetics transmitted through the popular mass media may become part of the buzz, but because its meaning and interpretation cannot be controlled, successfully employing this mode of transmission may entail significant challenges to the development of effective outreach.

The Influence of Structural and Cultural Forces

People become meaningfully engaged with information on genetics and health care as a result of life course events that require a greater understanding of genetics for making informed decisions for personal health care and family planning. These life course events, such as marriage, pregnancy, and aging are embedded within preexisting structural and cultural frameworks that influence the timing and degree of an individual's engagement with information on genetics and health care.

Biomedicine constitutes one such framework. Professional associations establish the guidelines and standards for prevention, diagnosis, and treatment of genetic disorders and conditions. These guidelines are published in popular media and incorporated into the relationship between primary health care providers and consumers, thereby influencing the timing, form, and content of information and services offered by providers to consumers. A number of studies included in this review demonstrate how providers control the flow of information and services to consumers (Friedman, et al. 1997; Hayflick, et al. 1998; Kluger, et al. 1991).

Furthermore, biomedical research on genetics results in new services and information that have the potential to alter the timing of individual engagement with this information. For example, much of the literature focuses on the "revolutionizing" effect that genomic research will have on medicine (Collins 1997; Collins and Bochm 1999; Fulginiti 1993; Kenner and Amlung 1999). Much of this literature is based on the assumption that a greater knowledge of the genetic contribution to disorders will result in an increased demand for tests that predict risk factors. "Gene chips" that provide personalized genetic profiles of individuals and their susceptibilities could indeed encourage consumers to engage information on genetics earlier, and in more meaningful ways (O'Hara

1999). According to some, however, the current provision of medical services and information has not been substantially altered by the advances in genomic research and resulting technologies (Holtzman and Marteau 2000; Wulfsberg 2000).

While research and practice in the biomedical field influences the timing and nature of consumer engagement with genetics and health, biomedicine itself is embedded in a larger political economy that structures the relationship between people and the health care system. Rayna Rapp provides poignant examples of how various factors, including language barriers, overcrowded clinic conditions, and low socioeconomic status may contribute to a late entry into genetic testing and counseling services (Rapp 1999:170-171). Such examples illustrate how barriers to access created by structural inequalities built into the health care system delay the timing of engagement for members of historically underserved populations.

The wider political economy is also affected by numerous competing interest groups that influence the direction of biomedical research and practice. For example, while research institutes, biotechnology firms, and private industry are creating more and more genetic tests for the market, the insurance industry is attempting to contain costs by refusing coverage for tests that they have classified as not beneficial and cost effective in the long term (Murray 1992; Reinhard 1996). Providers in managed care systems are often caught in a conflict of interests, facing the challenge of reducing costs while offering desired services such as genetics counseling for new genetic tests that may be costly or unavailable within their organization (Wingerson 1998:282).

In addition, private and government funding sources influence the direction of medical research, the technology that emerges from it, and the resulting guidelines established to incorporate it. Legislation governing such research is propelled by diverse interests and the variety of influences on biomedicine. For example, these influences include: efforts of the National Conference of Catholic Bishops to eliminate the public funding of embryonic stem cell research (Doerflinger 1999); the insurance industry's opposition to efforts aimed at limiting insurer access to genetic test results, and opposing attempts at lobbying to prevent insurance discrimination (Ad Hoc Committee on Genetic Testing Insurance Issues 1995; Aston 1997; Lowden and Roberts 1998; Rifkin 1998:162); and, lobbying by special interest groups, such as the Foundation on Economic Trends, for controls over genetics research (Rifkin 1998:63). All of these factors constitute the larger political economy within which biomedicine is embedded. They guide not only the information produced, but also the way such information is applied to the medical care of consumers.

While the structures and practices of biomedicine influence the timing of an individual's engagement with information on genetics and health, individuals, as consumers within the larger political economy, constitute a force that drives biomedicine. People do indeed bring information to the attention of their health care providers. This process is discussed in the health care literature as a significant factor compelling the need for increasing genetics competency among providers (Emery, et al. 1998; Kash, et al. 2000; Ludman, et al. 1999). The feedback between these related forces structures the timing of engagement with genetics and health care information.

Another dimension influencing the timing of engagement is the configuration of cultural beliefs, knowledge, and attitudes that guide decision making processes. Individuals and families are not part of an undifferentiated class of consumers. They are members of diverse sociocultural groups based on such features as: ethnicity; religion; geographic region; age; gender; and, socioeconomic status. These populations feature a variety of kinship and social relations, modes of communication, and historical experiences which together frame cultural value systems. Membership in socioculturally defined populations may shape attitudes toward biomedicine, mediate access to health care, and therefore influence the nature and timing of meaningful engagement with information on genetics and the health of individuals and families.

African Americans, for example, have many specific concerns about new genetic research and technologies. These concerns are related to the historical misuse of biomedical research in African American communities, including the Tuskegee syphilis experiment and the sickle-cell carrier-screening program (Jackson 2001). Focus groups conducted with African Americans as part of the "Communities of Color and Genetics Policy Project" highlighted a number of these concerns. Participants expressed distrust of the government due to an awareness of past abuses. African Americans also raised concerns about genetic profiling for health insurance and employment, and the need for privacy assurances. Skepticism over the goals of scientific research in general was discussed in terms of a perceived divide between scientific and spiritual values (Coe 2000). These findings suggest that culture-specific beliefs, values, and attitudes may influence the degree and timing of engagement with information on genetics and health care. Understanding the cultural characteristics of specific populations will help structure the creation of information and services in culturally appropriate ways. Coupled with the influence of the larger political economy on access to health care, sociocultural diversity becomes a very important factor influencing the timing of a person's engagement with information on genetics.

The influence of culture on the timing of engagement is illustrated by the establishment of *Dor Yeshurim*, a Tay-Sachs screening program for members of Hasidic and Orthodox Jewish communities derived from Ashkenazi populations

(Wingerson 1998). This program, endorsed by rabbinic authorities, is based on the concern that genetic testing for fetal abnormalities takes place too late in the life cycle. The testing is designed to aid in eliminating Tay-Sachs, and other deleterious genetic variations such as Gaucher disease, by forestalling marriages between carriers of these genetic variations. The program works by providing participants with confidential identification numbers that are assigned to their stored blood samples. Prior to marriage, culturally sanctioned matchmakers, parents, or donors themselves call the data bank and provide the identification numbers of the prospective spouses. If both blood samples referenced by these identification numbers test positive for Tay-Sachs, measures may be taken to find a more suitable match or to provide appropriate counseling. The success of this program is based, in part, on an intimate understanding of the role of religion and religious authority in the lives of Ashkenazi Jews. Educational media incorporates religious principles that resonate with the community. For example, a pamphlet produced by the program characterizes participation in *Dor Yeshurim* as religiously prescribed, or a "sacred duty" (Wingerson 1998:12). This example illuminates the power of culture to influence the timing of engagement with information on genetics and health care, and suggests the importance of understanding the dynamics of culture for the successful implementation of genetics literacy and genetic testing programs.

Life Events that Promote Engagement

There are many factors influencing the timing and degree of engagement with information on genetics and health care. Ambient genetics information gleaned on a day-to-day basis constitutes one form of engagement, the significance of which varies in accordance with personal life circumstances and events. More meaningful forms of engagement, precipitated by the life events discussed below, are mediated by the sociocultural framework within which they are embedded. While biomedicine dictates the standards that guide the development of timetables for assessing genetic health needs, biomedicine is influenced by the overarching political economy that drives research funding, produces legislation, shapes public perceptions, and structures access to health care. In addition, cultural knowledge, beliefs, and attitudes affect the timing and way in which genetics services and information are incorporated into individual and family practice. Viewing this system holistically provides an understanding of the complex, interrelated forces that shape a person's engagement with genetics and health care throughout the life cycle.

Reproductive Health

One of the primary life events for individuals and families that promote engagement with information on genetics and health is the anticipation and planning for childbirth. It is no exaggeration to say that reproductive events are no longer simple acts of nature. While family planning has a long history, more

recent developments such as the availability of birth control technologies, the changing workforce, the increased costs of having, raising, and educating children, and a host of other social and economic factors have all contributed to a widespread support for the rational planning of pregnancies, childbirth, and childrearing. New developments in reproductive technologies such as infertility treatments and *in vitro* fertilization have further expanded the opportunities for individuals and couples who could not otherwise have children. The emphasis on planning families is a significant cultural theme in some parts of modern American society, as evidenced by the stigma attached to unplanned or accidental pregnancies in some communities.

The biomedical establishment has endorsed this emphasis on planning through a variety of measures. Routine obstetric care encourages women and their partners to maintain good levels of health. Public health campaigns on fetal alcohol syndrome, folic acid deficiencies and neural tube defects, and the risks of smoking for pregnant women are prominent examples. In addition, interventions designed to limit teenage pregnancy and raise awareness concerning the risks of being sexually active at an early age tend to incorporate broader messages about the importance of rationally planning one's reproductive decisions. The overall message stresses that pregnancy should be, at a minimum, deliberate, healthy, and affordable. In short, planning families is a dominant theme, although not all groups or communities in our society accept all of its connotations, or all of the variations in message content.

New developments in genomic research bring an important new dimension to the emphasis on planning. On the one hand, as historical analyses of the family planning movement have demonstrated (Condit 1999a), certain kinds of genetics thinking and policymaking can easily be linked to various notions of eugenics. Current discussions about genetics and reproductive health often continue to resonate with these older concerns. It is no surprise that phrases such as "designer babies," "playing God," or "genetic enhancement" form part of the discourse regarding reproductive health. On the other hand, advances in genomic research may provide a deeper understanding of an individual's family and genetic history that may allow people to make more informed family planning decisions. The period in which an individual or a couple ultimately decides to become pregnant is crucial. It represents a time in which all couples or individuals, regardless of their current biomedical risk profile, may be contemplating what kind of person their child may become.

If the individual or couple has access to routine medical care and indicate that pregnancy is a possibility, the provider may refer them to a genetic counselor if they fit certain profiles based on criteria such as age, ethnicity, family history, or individual health history. However, as the literature suggests, the responsibilities of counseling and testing may increasingly fall on primary care providers, OBGYN

specialists, and other providers, given that genetics counselors are small in number and concentrated in major urban area.

Folk explanations of heredity, i.e., the grandfather's ears, the father's nose, the mother's smile, the uncle's temperament, become intertwined with fundamental concerns about the child's health and potential. Folk biologies and value systems vary between ethnic groups and populations. Rapp, for example, provides data that suggests that for some ethnic groups, such as Hispanics, appearing "normal" and thus avoiding stigmatization is intertwined with other notions of health (Rapp 1999). Furthermore, parental concerns about a child's genetic makeup, however they are conceived, are inevitably intertwined with their apprehensions and concerns about child rearing. Differing cultural notions about health, normalcy, childrearing and other reproductive issues represent challenges to developing culturally appropriate education materials. At the same time, they underscore the fact that the anticipation phase of child bearing and rearing represents a potential opportunity for education about genetics and health care.

Pregnancy has historically been the central life event during which individuals and couples become meaningfully engaged in genetics and health care. Early and regular prenatal care promotes the health of the mother and fetus. Initially, monitoring technologies such as ultrasound, chorionic villus sampling, amniocentesis, and genetic testing allowed providers to generate a relatively limited number of statements about the potential health outcomes of a fetus. Initially, the technology was valuable for detecting neural tube defects such as hydroencephaly and anencephaly and chromosomal disorders such Down syndrome. However, as genetic testing has improved, providers are now able to generate a substantial list of statistical and probability statements about the fetus's long-term health outcomes.

Even when statements about fetal health were limited, genetic counselors and providers faced a challenging task of translating those statements into useful information for an individual or couple. New tests and improvements in technologies have made the task even more formidable and costly. By necessity, health care providers will have to develop new ways of ensuring that individuals and couples can effectively interpret test results and make difficult decisions about continuing or terminating a pregnancy. As the literature review has suggested, there is considerable information available that may help individuals and couples make decisions. At the same time, the utility of much of that information depends on the level of genetics literacy of the individual or family. Furthermore, access to information is variable depending on the source of the information and the resources available to the individual or family.

Early Childhood and Pediatrics

The next stage at which individuals and families may encounter issues of genetics and health is early childhood. Pediatrics has had a long involvement in genetics and health care, both from the perspective of individual care, and from the public health perspective that emphasizes newborn screening programs. For new parents, newborn screening programs for PKU or congenital hypothyroidism may be their first encounters with genetic health care issues. However, given that these screening programs are designed to capture and direct parents of positive testing newborns into appropriate streams of care, parents of children who do not test positive may not perceive this event in terms of genetic health. At this time, we have not examined the types of supporting materials that parents receive regarding these screening programs.

The first years of development offer a series of potential encounters with genetics and health care. Currently, these encounters become meaningful engagements for the parents of children diagnosed with genetic disorders. The processes for channeling parents and children in this group are currently in place. In the next phases of the research process, these processes will be explored in greater detail. The current consumer organizations who are partners in this research project play a vital role in providing support and educational materials. The literature also suggests that additional outreach and coordination of effort is desirable, especially in light of the changes in technology that will make additional testing services available without addressing issues of access to services.

The monitoring of early childhood development, as with amniocentesis and screening efforts, are designed to detect health problems. In the case of infants, the detection of developmental delays in motor, language, and cognitive skills may generate a series of diagnostic procedures that are highly complex. At this stage, providers must diagnose and assess the effects of environmental, behavioral, and genetic factors. Depending on the nature of the specific disorder, these events may produce high levels of anxiety for parents. As research into the underlying genetic components of developmental processes and potential disorders progresses, providers will have more powerful diagnostic tools that will necessitate addressing the challenges of communicating information regarding the etiologies, treatments, and prognoses of disorders. And, as some parents become more aware of genetics issues, simple explanations that emphasize a particular cause may not be sufficient. Issues of diversity and access complicate the communication process and consequently may compromise standards of care across populations. Changing demographics within the United States further underscores the importance of developing supporting programs that are of high quality yet flexible enough to address issues of cultural diversity.

Adolescence

Children mature and enter the formal education systems where their motor skills, social skills, and learning patterns are increasingly monitored. This review focused on more narrow definitions of health issues, and has not currently addressed the extensive literature on education, learning disabilities, and special education. Nevertheless, this subject warrants mention given its historical relationship to larger themes that have involved genetics issues. For example the extensive literature on IQ and "intelligence testing" vary widely. Discussions about whether or not tests measure "innate" or heritable abilities have been highly charged, given that many studies have attempted to measure differences between and among ethnic groups. These discussions are part of a larger discourse on behavioral genetics, a field that is growing, prolific, and aggressive in its willingness use simple Darwinian selection arguments to explain social and behavioral variation (Harrington 1997). The more extensive social critiques that have investigated the impact of genetic research place this type of literature within broader historical trends that are linked to the eugenics movements of the early twentieth century (Allen 1997; Nelkin 1995).

Standardized testing and monitoring will no doubt remain a central feature of the American educational system, and portions of the social scientific community remain powerfully committed to research that investigates the genetic bases of behavior, "intelligence," "violence," and other complex behaviors. While this project focuses more narrowly on genetics and health care issues, studies on the genetic basis of behavior raise the decibel levels of the buzz considerably (Rifkin 1999). Any toolkit that promotes genetics literacy would presumably not have to focus on these issues exclusively, but it must address them at some level if it hopes to have buy-in from those communities that are concerned about access to good health care and good education, as well as fair and just treatment.

Young Adulthood to Middle Age

Young adulthood to a large extent provides the basis for definitions of adult healthfulness. Protocols for preventing disorders that may present during adulthood such as cardiovascular disease, obesity, diabetes, or asthma call for basic early monitoring of diet, behavior, lipids, and cholesterol profiles, especially for individuals with positive family histories. It seems likely that at this time, providers and consumers do not perceive these basic tests as genetic in nature. Nevertheless, as ongoing research makes it possible to more precisely determine the genetic components of disorders and the genetic basis of risk for these disorders, such monitoring becomes more genetic in character.

It is important to emphasize here that monitoring protocols differ greatly by gender. The most prominent examples in the literature focus on determining genetic factors for cancer, with breast cancer receiving the most attention. The discoveries of mutations on the tumor suppressor genes BRCA1 and BRCA2, the

subsequent development of tests for mutations, and the difficulties in integrating these findings with environmental factors suggest that research and testing are still in the early phases of development (Gilliland 1997; Johnson, et al. 1997). The literature highlights the problems of communicating information about risk in light of these developments to consumers in meaningful, understandable, and culturally appropriate ways (American Academy of Family Physicians 2000; Browne and Romilly-Harper 2000; Davis 1997; Durfy, et al. 1998; Geller, et al. 1997b). In addition to monitoring and testing for breast and ovarian cancer, tests are being developed for prostate cancer in males. Presumably, tests for other forms of cancer will emerge as research progresses. The American Society for Clinical Oncology has already developed guidelines for developing educational and curricular materials related to genetic testing for cancer (American Society of Clinical Oncology 1997b).

In sum, the new tests for colorectal cancer and breast cancer offer medical providers and consumers new tools for identifying "at-risk" individuals. In turn, these tools may enhance monitoring for early detection and treatment. However, the literature emphasizes that failures to properly communicate information about risk have potentially damaging psychological effects. Such challenges also raise ethical issues given that genetic information about a single individual is by its nature extended to related individuals who may or may not wish to receive information about their own risk profiles. These findings underscore the importance of developing information that is sensitive to ethical issues and designed to take into account cultural conceptions of relatedness.

Current Issues Related to Aging

Recent research into the potential genetic factors of Alzheimer's disease highlight how issues related to developments in genetic and health care affect late stages of the life course. As people live longer and older consumers comprise a greater portion of the overall population, issues of aging, appropriate health care, health care costs, and the quality of life for older consumers will continue to gain prominence. Alzheimer's disease and forms of dementia pose a particular challenge for our health care system. Individuals with Alzheimer's disease may be physically healthy and lead long lives while simultaneously requiring high levels of care.

The adult children of parents with Alzheimer's disease or other kinds of chronic disorders will bear a disproportionate burden of managing their parents' care and quality of life. In this context, new tests for protein markers that indicate the likelihood of early onset Alzheimer's disease, or the APOE test that measures risk factors for sporadic late-onset Alzheimer's, prove most useful, if administered in ways that promote planning for long term care (Panegyres, et al. 2000). Currently there are questions about the predictive value and ethical implications of these tests (Fleck 1998; Kahn 1997; Panegyres, et al. 2000). This literature

urges caution and deliberation in the implementation of these tests. These concerns will also apply as development in genetic sciences and biomedicine reveal genetic bases to other disorders and conditions associated with aging.

Conclusion: Life Events and the Development of Effective Outreach

Using the life cycle to assess when consumers' encounters with genetics and health care are translated into meaningful engagements with information allows the discussion about genetics and health care to be situated within the larger biomedical monitoring systems that are currently in place. Furthermore, this analysis suggests that certain stages in the life cycle, such as when individuals and couples are anticipating and planning to have children, are moments when consumers' desires for information on genetics and health care may be greatest and when they may begin to actively seek such information.

Encounters with genetics information continues as an individual passes through different life stages. However, risk profiling and monitoring for many disorders and conditions are for the most part not viewed as genetic health issues by individuals and families. Rather, they represent potential encounters in which consumers may become meaningfully engaged in genetics and health care issues provided that the monitoring process is framed in a way that captures the interactions of genetic, environmental, cultural, and behavioral factors.

At the same time, genomic research and developments in biotechnology also portend a radically new approach to health care, where genetic profiling at conception or prior to conception generates a wealth of information about the health and long-term viability of an individual, all stated in the mathematical language of probabilities and risks. However, there are strong tendencies to translate open-ended indeterminate risk statements into statements about whether risk for disorders equates with heritability. This is a process already in place, since insurance providers must collapse complex statistical realities with indeterminate outcomes into manageable actuarial units for purposes of cost accounting. In that regard, statistical risk profiles have a real effect on access to care and costs associated with care.

The potential front-loading of information on one's personal genetic make-up has sparked serious moral and ethical debates about the degree to which such information is relevant for improving health outcomes. Furthermore, the potential for front-loading has raised concerns about the ways in which some parties may use such information to discriminate against individuals or groups. If this potential for front-loading is coupled with experiments in genetic engineering, pre-implantation diagnoses, and the selective termination of pregnancies, serious concerns are raised about the potential abuse of medical

technologies and procedures for non-medical or "enhancement" purposes or even market driven eugenics. The standards and guidelines developed by the biomedical establishment will no doubt undergo serious transformations as genetics is integrated into biomedicine. At this time, however, it is difficult to determine the pace of those developments. It is worth reiterating that the societal buzz regarding genetics and health care is generated by diverse forces, including private sector companies that are developing and marketing new genetic tests. While the biomedical establishment continues to develop standards and guidelines, actual medical practice does not adhere perfectly to these guidelines, and consumer demands for new kinds of genetic testing will no doubt influence the future of these developments. In essence, these developments are consumer driven, so as individuals and families are presented with new opportunities to engage meaningfully with information about genetics and their health, outreach initiatives must develop with an understanding of the broader social implications of advancements in genetics-based medical care.

Health Seeking Behavior

Question 3: How do people find information on health care and genetic health care in particular?

The three guiding questions in this research phase have been oriented toward understanding the processes by which information about genetics and health is generated, disseminated, accessed, and assimilated. The first two questions focused on the larger organizations and structures associated with genomic research. This final question explores the way in which people seek and find information on genetics and health care. The review of the literature suggests that this subject has not been systematically addressed. The educational initiatives directed towards health providers assume that consumers will learn about genetics in the traditional health care environment. The major social critiques that explore public attitudes and perceptions of genetics and health care rely primarily on textual analyses in order to assess ongoing historical trends. Content analyses of science writing and journalism also provide suggestions about how consumers are accessing and making sense of information about genetics and health care. These approaches allow us to make inferences about consumers' knowledge, attitudes, and values, but they provide little direct evidence for the ways in which people actively seek information on genetics and health care. This final question provides a framework for addressing the gaps in knowledge that drive the future phases of research into decision making among current consumers. The gaps are illustrated by a brief review of the consumer research literature.

Lessons from Consumer Research

The review of consumer research did not reveal any systematic attempts to determine how people find information specific to genetics and health. A number of studies, however, provide insights into how people receive and seek information on health care in general. Most of the information emerging from this research is based on quantitative surveys and telephone interviews that may not fully capture the processes of health care decision making, nor the sociocultural diversity underlying them. What people say they do regarding the search for health care information may diverge significantly from the process they actually follow. Furthermore, this process varies in accordance with cultural knowledge, beliefs, and attitudes, and is mediated by issues of access to information and services. Nonetheless, the studies offer valuable insights into stated preferences that, when coupled with qualitative analysis of actual practice, may guide future stages of research in support of developing effective outreach.

A recent study conducted by the Pew Center is an example of consumer research that illustrates dimensions of health care decision making (Fox and Rainie 2000). The Pew Internet and American Life Project (www.pewinternet.org) found that 55 percent of Americans with Internet access use web searches to find information on health care. According to the study, the majority of these "health seekers" use the Internet at least once a month to learn about health-related issues for themselves or on behalf of a family member, companion, or friend. The vast majority (91 percent) of "health seekers" use the Internet to find information on physical illnesses, and most respondents reported that this was in conjunction with a doctor's visit (61 percent). Seventy percent of the "health seekers" claimed that the information they received influenced the way in which they treated illnesses or conditions. While "health seekers" enjoy the convenience of the Internet for accessing health care information, 85 percent expressed concern about privacy violations and the possibility that insurance companies might deny coverage to those who visit health care sites.

The study offers many other valuable findings, including demographic features of the sample and notable gender differences. Interestingly, however, the study does not capture any sociocultural differences in decision making. According to the authors, "the seeking of health information (on the Internet) is equally compelling to all racial and ethnic groups. Similarly, there is no major 'income' effect on this activity" (Fox and Rainie 2000:9). However, these findings are relevant for assessing the importance of the Internet as a tool for acquiring health information only for those who have access to the Internet. There may be significant variation in access to the Internet based on race, ethnicity, and income. Furthermore, because the study was not designed to elicit the health care decision making processes, intercultural variation in such practices was not considered. In short, the study may overstate the potential importance of the Internet in the "health seeking" behavior of those who either do not have Internet access, or who do not actively seek health care information on the Internet despite having access. For example, a 1998 survey conducted by Princeton Survey Research Associates found that 66 percent of adult respondents received no health information from the Internet (Princeton Survey Research Associates 1998). The same survey found that, among African Americans, much more information is received through places of worship than the Internet. The comparison of these surveys illustrates the need to contextualize the search for information on health care within the sociocultural framework within which access to information is regulated.

A qualitative study conducted by Balch Associates on perceptions of genetics research supports the finding that people receive the most information on genetics through mass media channels, including television, newspapers, and radio (Balch Associates 1999). The study reported that only some people found information on genetics through the Internet. The study also found that, with

the exception of those who have a genetic disorder themselves or in their family, people do not actively seek information on genetics. However, the study was careful to point out that people do not categorize research on genetic disorders as "genetics research." People in general do not have a cognitive category within which genetics research is compartmentalized and organized. In other words, genetic disorders are treated no differently than other illnesses, suggesting that seeking and finding information on the relationship between genetics and health care may be a by-product of health seeking behavior in general by individuals or families.

If the receipt of information on genetics and health care is, in fact, an epiphenomenon of broader health seeking behavior patterns, an analysis of the health seeking process may illuminate the ways in which people come to learn about genetics. Examining health seeking behavior also highlights the necessity of attending to sociocultural differences by illuminating the influence of social history, cultural knowledge, beliefs, and attitudes on medical practices. Finally, considering the "hierarchies of resort" in medical decision making by individuals, reveals the ways in which information related to genetic health differs from other health information, and highlights the particular challenges raised by these differences.

Health Seeking Behavior

Anthropological research on medical decision making has developed a number of flexible and generalizable models that allow researchers to capture and compare health systems across space and time (Freidenberg 2000; Hellman 1984; Kleinman 1980). Kleinman's "Explanatory Model of the Health Seeking Process" divides the process into four different steps: 1) perceiving and experiencing symptoms; 2) labeling and valuating disorders and conditions; 3) sanctioning a particular kind of sick role; and, 4) deciding what to do and engaging in specific health care service. The steps make no assumptions about the system of health care delivery. Rather they assume that while illness is a cultural universal, the expressions of symptoms, the diagnostic process, and the ways in which illnesses are treated are all ultimately culture bound (Lock 1993; Zhang, et al. 1998). This assumption has profound implications for the ways in which providers who are trained in and accept a biomedical worldview interact with individuals who come from divergent cultures. The literature on cultural competence and cultural sensitivity suggests that the biomedical establishment recognizes that providing good care requires the ability to appreciate language and cultural differences, if only to improve the communication process. The bioethics literature that emphasizes informed consent and non-directive counseling affirms this approach more strongly. It recognizes that even when the cultural differences between the provider and consumer are minimal, the provider is obligated to respect the consumer's value system and autonomy.

The American health care system is dominated by biomedicine, but it is still a plurality of co-existing modes of care. Folk systems and a growing number of holistic medical systems derived from folk or traditional perspectives may all compete with or complement the biomedical approach. Within that framework, it is necessary to consider the practical issues of access that further structure consumer decisions about genetics and health care. For example, a lack of health insurance, language barriers, or other socioeconomic factors may compromise an individual's access to good prenatal care, or to the kinds of testing and monitoring that trigger encounters with genetics and health care in general. In addition, not all ethnic groups or communities have had the same historical relationships with the dominant biomedical health care establishment. Individuals may have practical access to biomedical health care, but their lack of trust or mistrust in the system may affect how they seek care (Millet and Khanani 1997). Finally, some groups or individuals may simply choose not to participate in the mainstream biomedical system. In some instances, these choices are constrained by law, but in others, they are left to a negotiation process between those individuals who may be wavering, and the providers who are obligated to provide the best course of treatment.

Within this structural context, Kleinman's model applies to all disorders and conditions as they are detected, diagnosed, and treated. However, medical advances based on genomic research provide information about health that may alter how the basic model operates as an individual makes medical decisions. It is important to consider the stages in medical decision making in light of these advances.

Perceiving and Experiencing Symptoms

The process by which an individual perceives and experiences symptoms of a disorder or condition varies within and between groups and populations. Basic experiences of pain, hotness, coldness, internal discomfort, malaise, or fatigue are subjective. The individual uses the folk models they have learned to perceive those basic experiences both in degree and in kind. An individual may perceive neck pain as a type of "stress" headache; back pain as a kind of injury; or, malaise as a kind of fatigue or sadness. In turn the individual may perceive these symptoms as mild and trivial, or as severe enough to warrant seeking treatment.

Many culturally derived assumptions tend to link illness with the appearance of symptoms. Information derived from genetic testing has the potential to change this first step in health seeking behavior, though culturally derived assumptions about illness may present a significant barrier to changing health seeking behaviors. Genetic testing is often used for predicting future disorders. At the point when an individual receives information about genetic predispositions, she or he may not experience symptoms and place them in a folk model of illness.

To fully benefit from predictive diagnoses in the biomedical model, an individual should seek preventive care before symptoms emerge and illness is defined. Consequently, the consumer may suspend preventive measures until such symptoms occur which are associated with a disorder that would lead them to seek care. For many populations, there is a reluctance to engage with the health care community unless a condition is emergent and marked by symptoms.

Labeling and Valuating Disorders and Conditions

Disorders and conditions are usually labeled and given a culturally defined value after an individual experiences symptoms that indicate a non-normative state of health. This process involves assigning a label to the symptoms, determining the severity of the disorder, and defining the implications that the disorder has for the affected individual and society as a whole. In contemporary American society, there is a plurality of medical and cultural systems that shape this process. Cultural definitions of the body, assumptions about illness and health, and an entire constellation of values are embedded in medical models. Varying ethnic valuations of disorders such as Down syndrome illustrate the complexity and variability underlying women's reproductive decisions following positive screening (Rapp 1999). These models vary among ethnic populations and although they may conflict with biomedical models, they may incorporate important elements from biomedicine. Access to biomedicine, defined in sociohistorical and economic terms, often determines the extent to which people rely on alternative models. How people participate in this second stage also affects the way in which people seek and acquire information on genetics and health care.

The literature on how people acquire information on genetics and health care suggests that people intensify their explorations only after the second stage of the model. In other words, once a person's condition has been diagnosed, labeled, and valuated as a "genetic" disorder, they or their loved ones begin searching more specifically for information on genetics and health. How they intensify their search and where they go for the information will be explored in future stages of research. The intensification of efforts may be due to the fact that genetic disorders are seen as having greater psychosocial implications for present and future family members due to the hereditary nature of genetic disorders. It may also result from an acceptance of the essentialization entailed by labeling a disorder "genetic." Genetic essentialization may be exacerbated by new genetic technology that creates labels for future states of disorders before symptoms are experienced. This may have important implications for the cultural definition and valuation of disorders. It may also transform the timing and method of health seeking behavior.

Understanding cultural variability in the medical labeling and valuation of disorders and conditions will guide the development of culturally appropriate

outreach efforts. For example, research on this stage suggests that the acceptance of biomedical definitions and labels is often dependent on the establishment of community-based education efforts. As Rapp points out, groups for parents with disabled children provide support and information that normalize biomedical definitions of genetic conditions (Rapp 1999:302). Understanding this process may guide future efforts to demonstrate the value of genetics literacy in culturally appropriate and meaningful ways. For example, the underutilization of genetics services by minority communities motivated the development of a pilot project designed to educate African Americans and Russian immigrants in Baltimore, Maryland (Mittman 1998). The project's success, according to the author, was directly related to the incorporation of cultural values and the successful use of existing social support networks in the creation and implementation of genetics and health care education programs.

Sanctioning a Sick Role

For this step in medical decision making, individuals must assume a social role or roles that are appropriate for a sick person. These roles vary cross-culturally and within cultures. Societies that rely on folk remedies may at one extreme sanction social isolation or even banishment for individuals with contagious or stigmatizing disorders. At the other end, they may incorporate individuals with disorders into culturally sanctioned roles that provide ongoing social support and security.

The biomedical approach typically follows a formal procedure that sanctions and integrates the ill person into a series of procedures that focus on monitoring and treatment. For biomedicine, illness is a matter best left to experts and formal systems. Social support networks in this system tend to be viewed as adjunct to the formal system, and are seen as useful in encouraging compliance with monitoring and treatment. Once again, these procedures are constrained by questions of access and varying beliefs systems. Under the formal system, acute disorders motivate consumers to seek out providers until the afflicted individual's health returns to normal. After that, the individual interacts with the system via regular monitoring regimes that focus on prevention of symptoms. For chronic disorders, the individual develops ongoing relationships with providers. In these relationships providers monitor the progress of the disorder, adjust treatment regimes, and monitor behavioral elements. The goal is to control the disorder so that the individual can continue to lead a full lifestyle. In recent decades the biomedical approach has incorporated initiatives designed to empower the individual and give them a greater role in managing their disorder. These initiatives assume that individuals will be able to monitor their own disorders and comply with a regime designed to control the progression of the disorder. When utilized effectively, these initiatives reduce health care costs and allow individuals greater flexibility and autonomy.

The information generated by advances in new genetic technologies has powerful implications for monitoring these two different kinds of disorders. In short, new information about an individual's genetic predispositions for both acute and chronic disorders necessitates more intensive health monitoring. For those disorders that are known to be deeply penetrant, monitoring will capture symptoms as they emerge. For chronic disorders, informed individuals, in conjunction with their providers, will be able to more closely monitor environmental and behavioral components.

On another level, genomic research also holds the promise of developing new drugs and treatments, such as gene transfer therapy, that in effect render both acute and chronic disorders more manageable. It is conceivable then that new genetic technology can radically redefine illness and health. Formerly acute but untreatable disorders may become chronic manageable ones. Society may no longer view individuals with chronic disorders as being "sick" given that their disorders are manageable. At this time however these types of treatments are in the early experimental stages, and are progressing more slowly than the ability to detect the presence of traits using genetic testing. The lag time between diagnosis and treatment presents potential problems regarding how people respond to genetic information.

On the other hand, genetic diagnoses that precede the experience of symptoms may encourage radical interventions that are not warranted in consideration of biomedicine's ability to assess risk. The decision to undergo a "prophylactic mastectomy" after testing positive for BRCA1 or BRCA2 genes is an example of how genetic technology can encourage the use of radical treatment for a pre-diseased state (Matloff, et al. 2000; Rothman 1998:151; Saunders, et al. 1999). Assigning a diseased state through predisposition testing may also contribute to a sense of fatalism that entails significant psychological distress (Kash, et al. 2000; Press, et al. 2000). Finally, negative test results may sanction behavior that increases risks for common disorders such as cardiovascular disease if such results are perceived as definitive statements on one's future health.

Engaging in a Specific Health Care Service

This model suggests that people usually begin to seek health care information when they experience symptoms. The initial step in the information search is the labeling and valuating of the disorder. This process of labeling, sanctioning a sick role, and engaging in a specific health care service is culturally variable and dependent on people's access to different types of information and services.

In this final stage, individuals become consumers of a variety of health care services that may include folk healers, alternative medicine, self treatment with over the counter medicines, or biomedicine. Access to biomedical treatment in part structures consumers' choices for particular kinds of health care services.

Those consumers who access biomedical care episodically may not incorporate biomedical explanations of disorders to the same degree as those who have greater access to biomedical services and information. Groups and individuals who have less access to biomedical services consequently may not accept and incorporate information on genetics and health into their health care decisions.

In the biomedical model, the monitoring of health indicators such as weight, cholesterol levels, activity levels, diet, or environmental factors, that are part of routine care, already provide risk information about future health outcomes prior to the experience of any symptoms. This type of monitoring already allows the individual consumer to label and valuate a disorder prior to the experience of symptoms. In this respect, the new technologies associated with genetic testing are not qualitatively different from other kinds of preventive monitoring.

Within this framework of prevention, consumers are encouraged to engage in biomedical health care services on an ongoing basis for purposes of health promotion and disease prevention. Yet, while this may be seen as an ideal model for the future, current medical practices do not include routine genetic testing as part of their monitoring regimes. Currently, less invasive and less costly procedures such as the taking of family histories, newborn screening, and amniocentesis provide the bulk of data related directly to the monitoring for genetic conditions and disorders. As such, these current tests and screening systems are key moments when consumers have the opportunity to incorporate information on genetics and health care into their decision making.

This has important implications for subsequent research stages in this project. Biomedical care channels consumers and their families into standardized treatment modalities. Parents of children with positive test outcomes are encouraged to learn about their children's disorders, and to seek support in their child's treatment. Individuals who test positive at a later age or in adulthood are also encouraged to learn about their disorder and treatment. Currently, there are a wide variety of disorder-based groups that function to provide support and information for parents and individuals after a diagnosis. In addition, a variety of religious, ethnic, and community-based organizations exist that may provide social support as well as information about genetics and health care. Research that identifies these groups and organizations and the kinds of services and information they currently provide is an essential step in developing effective outreach.

At each of these stages, individuals and families seek information about genetics and health. Understanding how individuals seek information differently at each stage is crucial to understanding medical decision making. These differences also affect the choices people make regarding where to seek and ultimately find useful information. As the research progresses, questions about the medical

decision making processes that people use and the sources they turn to for information will be addressed.

Conclusion: Lessons Learned

In this report the materials have been organized into five categories of information production: Primary Genomic Research and Biotechnology; Medical Provision and Treatment; Social Science and Policy Analysis; Community Education and Outreach Initiatives; and, Science Journalism. These categories capture the macro-processes by which the raw data of primary genetics research is translated into information valuable to experts in related fields, and then is ultimately transformed and disseminated as knowledge that may or may not help consumers make informed decisions about their health care.

This review raises several issues that should be addressed in further research and offers valuable lessons for guiding next steps in the research process and developing effective outreach. There are lessons learned that are essential for understanding how information about genetics and health is communicated to the public. Lessons regarding cultural appropriateness are also highlighted as guides for future stages of research and the development of the outreach content and strategies. In addition, outreach strategies must be designed to take into account timing issues that drive consumers to seek information on genetics and health in response to key life events and to make health care decisions related to genetics. These categories of lessons learned are bulleted separately below as indicators to guide continued research and as markers for the development of strategies and messages.

Issues Regarding Communication to the Public

- The social science and policy analysis literature provides an important context for understanding the broad implications of genetic research. At the same time, however, it is difficult to assess the degree to which the analyses, proposals, and solutions offered in this literature has influenced different sectors of the public in terms of decisions about their health and health care choices.

- Information is generated by private corporations, not-for-profit organizations, and the public sector. From the consumer perspective, however, information found in the private sector is not necessarily differentiated from basic research findings in the public sector and academic settings. Rather, public perceptions of developments in genetics relevant to health combine information from all of these sources.

- Much of the information being generated with ELSI funds is communicated within circles of health care providers and academics. There does not appear to be any systematic effort to translate or package information from ELSI-funded studies specifically for consumers either as targeted efforts to improve genetics literacy or in the form of community outreach programs based on public health approaches. Information on the ethical, legal, and social implications of developments in genetics and health care must be incorporated into effective outreach strategies and messages.

- The Internet is one of the primary channels for disseminating technical information. Virtually every research institute and biotechnology firm that focuses on primary research maintains Internet databases and websites for ease of access and rapid communication. An outgrowth of this use of technical information has been a variety of sites and portals that provide information about genetics and health.

- Journalism and science reporting tends to treat the audience as undifferentiated, individual consumers, glossing over the many issues of relevance to underserved populations.

- While scientists are often dissatisfied with the particular topics or modes of translation that journalists use, they must also rely upon science writers to publicize their work. The translation may not be effective, but this feedback loop, from research to popular media, is one of the primary means whereby information about genetics and health reach the public.

- Information on genetics transmitted through the popular mass media may become part of the buzz, but because its meaning and interpretation cannot be controlled, successfully employing this mode of transmission may entail significant challenges to the development of effective outreach. Participants in the creation of information on genetics and health care must be aware of the power of language to influence public perception, and accept the burden of responsible reporting.

- According to a focus group study conducted for the National Health Council, journalistic mass media is the primary mode through which members of the general public receive information on genetics and health care (Balch 1999). Other studies suggest that clinical office visits are one of the primary means by which providers communicate information on genetics and health care to consumers. Further research into how consumers receive information about health and

genetics and their knowledge of and attitudes toward health and genetics is necessary.

Issues Regarding Cultural Appropriateness in Effective Outreach

- Programs or interventions that define increased knowledge strictly in a biomedical sense without taking issues of cultural diversity into account may not result in a heightened consumer interest and confidence in genetics services.

- Primary care providers assume the role of gatekeepers to the world of genetics and health care, providing primary education for consumers according to a prescriptive approach to care. Health care researchers and providers have made few systematic attempts to specifically educate consumers. Understanding cultural variability in the medical labeling and valuation of disorders and conditions will guide the development of culturally appropriate outreach efforts.

- The importance of establishing a toolkit for genetics literacy is supported by the genetics counseling literature, much of which demonstrates a concern for the psychosocial and cultural context within which genetics information and services are provided.

- The public health literature offers many insights into understanding how genetic research will affect populations and communities. It is particularly important in that it emphasizes environmental and developmental issues related to genetics and health care.

- The literature that integrates the insights of social science into bioethics approaches reinforces the importance of cultural appropriateness by placing genetics health care within a broader community outreach and public health framework. It further emphasizes the importance of equal access to genetics information and services for all consumers.

- Ethnic groups and communities provide the social structure through which culturally appropriate research and outreach associated with genetic disorders may be conducted. The aim is not to link genetic disorders to ethnic groups, but rather to educate individuals about potential genetic risks that they <u>may</u> share within groups or communities that have increased risks for certain kinds of disorders due to genotypic penetration into the population, and to do so in an appropriate way.

Issues Related to Timing of Effective Outreach

- People encounter information about genetics and health care on a daily basis, but the degree to which they engage this information varies in accordance with a host of personal life events, circumstances, and external forces.

- Certain stages in the life cycle such as when individuals and couples are anticipating and planning to have children are moments when consumers' desires for information on genetics and health care may be greatest and when they may begin to actively seek such information.

- At each stage in the health care seeking model, individuals and families seek information about genetics and health. Understanding how individuals seek information differently at each stage is crucial to understanding medical decision making. These differences also affect the choices people make regarding where to seek and ultimately find useful information. As the research progresses, questions about the medical decision making processes that people use and the sources they turn to for information will be addressed.

- The developing and marketing of new genetic tests is consumer focused. Individuals and families are presented with new opportunities to engage meaningfully with information about genetics and their health based on these developments. Outreach initiatives should be developed with an understanding of the broader social implications of advancements in genetics-based medical care that have an impact on the health care choices faced by individuals and families.

A Summary of Guiding Questions for Further Research

The issues detailed in this report provide significant lessons learned which also raise important questions for the ongoing research process in support of the development of effective genetics education outreach. They reflect the three central questions that guided the research to this stage, and they extend our understanding of the current state of information production and dissemination regarding genetics and health. These questions will continue to guide the research, and they have raised new and interesting issues which must be considered as the research evolves. These include, but are not limited to:

- What does the general public know and understand about genetics?

- Through what channels does the general public obtain information?

- What is the minimum level of knowledge that the general public requires in order to make informed decisions about their health as new developments in genomic research and biotechnology change the biomedical view of health and the provision of medical care?

- And, how can this information be transmitted most effectively?

As the research evolves, these questions will be considered within a variety of populations and are expected to be refined and restated. As discovery continues, the materials and review process will be ongoing as a central component of continued research.

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Appendix 1:

Glossary of Selected Terms

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Achondroplasia: An inherited genetic disorder that affects bone growth, as manifested in "dwarfism."

Amniocentesis: A procedure usually performed between the sixteenth and twentieth week of gestation for obtaining amniotic fluid used in screening for fetal genetic abnormalities.

Ashkenazim: Ethnic populations of Eastern European Jewish descent.

Autosomal Recessive: A genetic condition that appears only in individuals who have received two copies of a gene located on a nonsex chromosome.

Bioinformatics: The integration of computer technology, information sciences, mathematics, and molecular biology in human genomic research.

Chorionic Villus Sampling (CVS): Procedure used between the eighth and tenth week of pregnancy in which tissue from the placenta is excised and used for prenatal diagnosis.

Congenital Adrenal Hyperplasia: A genetic disorder that impairs the adrenal cortex with subsequent overproduction of adrenal androgens.

Congenital Hypothyroidism: A genetic disorder characterized by a reduction in the activity of the thyroid gland resulting in developmental delays.

Cystic Fibrosis (CF): A genetic disorder affecting the exocrine glands that is characterized by the production of abnormal secretions, leading to mucous build-up that can impair pancreas, lung, and intestine function.

Down Syndrome: A common chromosome disorder resulting from an extra chromosome number 21 (trisomy 21) that affects physical and mental development. It is associated with increased risk for heart malformations.

Folic Acid: One of the B vitamins that is a key factor in the synthesis of nucleic acid (DNA and RNA). Lack of adequate folic acid intake during pregnancy increases the risk of congenital heart malformations, cleft lip and palate, urinary tract defects, and birth defects involving the spinal cord and brain

(neural tube defects), such as spina bifida (meningomyelocele) and anencephaly.

Fragile X Syndrome: The most common heritable form of mental retardation caused by a genetic mutation on the X chromosome.

Gaucher Disease: A progressive genetic disorder resulting in enlargement of the spleen, low red blood cell counts (anemia), a decrease in blood clotting cells (platelets), increased pigmentation of the skin, and a yellow fatty spot on the white of the eye (a pinguecula). It is most common in Ashkenazi Jewish populations and is the most common genetic disorder among Jews in the United States.

Gene Transfer Therapy: The treatment of a genetic disorder by replacing, altering, or supplementing a gene that is absent or abnormal.

Genomics: The study of the structure and function of the genome or, the entirety of genetic material in the chromosomes of a particular organism.

Genotypic Prevention: Medical interventions designed to reduce or prevent the transmission of particular genotypes to future generations.

Hemochromatosis: A genetic disorder resulting in the buildup of excess iron in the body that damages the liver and other organs.

Huntington Disease (also Huntington's Chorea): A genetic disorder characterized as an "adult-onset disease" resulting in mental and physical deterioration leading to death. The gene associated with the disorder contains a trinucleotide repeat - an unstable repeating sequence of 3 nucleotide bases (CAG) in the DNA.

Marfan Syndrome: A genetic disorder of connective tissue characterized by long fingers and toes, visual impairment, aortic wall weakness, and aneurysm.

Maternal Serum Alpha-fetoprotein (MSAFP): A plasma protein in maternal blood normally produced by the fetus that is used in screening for neural tube defects (anencephaly and spina bifida), Down syndrome, and other chromosome abnormalities.

Microcephaly: A condition marked by an abnormally small skull caused by a wide variety of problems that impair brain development including infections, radiation, medications, chromosome abnormalities and genetic disorders.

Monogenic Disorder: A genetic disorder caused by a mutation at a single genetic locus such as Huntington disease, cystic fibrosis, and sickle-cell disease.

Neurofibromatosis: A genetic disorder characterized by "cafe-au-lait" (light brown) spots on the skin, and a tendency to develop large tumors on the nerves, skin, and internal organs.

Neural Tube Defect (NTD): A birth defect caused by abnormal development of the neural tube, the embryonic structure that gives rise to the central nervous system. The best-known neural tube defects are anencephaly (absence of the cranial vault and absence of most or all of the cerebral hemispheres of the brain) and spina bifida (an opening in the vertebral column protecting the spinal cord).

New Genetics: The constellation of emergent technologies, research practices, information, and services related to genetics that is built on the integration of diverse fields including bioinformatics, proteomics, genomics, medicine, and industry.

Pharmacogenomics: The study of the interaction between drug interventions and molecular biology.

Phenotypic Prevention: Medical interventions designed to forestall and/or treat the physical expression of a genetic disorder.

Phenylketonuria (PKU): A genetic disorder marked by the inability to process the amino acid phenylalanine that may lead to mental impairment.

Polygenic Disorder: A genetic disorder caused by the interactions between or mutations in more than one genetic locus.

Pre-implantation Diagnosis: A procedure performed on DNA extracted from an embryo that has between four and sixteen cell divisions that is designed to identify chromosomal abnormalities.

Proteomics: The study of the proteome or, the entirety of proteins produced by a particular organism. Proteomics is concerned with understanding the molecular structure of proteins, their genetic basis, and their biochemical and physiological functions.

Recessive Ataxia: A genetic disorder characterized by muscular degeneration that is found in higher frequencies among French Canadian and Cajun populations.

Rett Syndrome: A genetic disorder characterized by impaired neurological development that is one of the most common causes of mental retardation in females.

Sickle-Cell Disease: A genetic blood disorder characterized by abnormal hemoglobin that deforms red blood cells, impairing blood and oxygen transfer to organs. It is particularly common among populations of African and Mediterranean descent.

Stem Cell Research: Research on that class of relatively undifferentiated cells that retain the ability to divide throughout life and produce specialized cells. This research often focuses on human embryonic stem cells: cells derived from human embryos or human fetal tissue that are self-replicating and known to develop into cells and tissues of the three primary germ layers.

Tay-Sachs Disease (TSD): A genetic disorder characterized by an enzyme deficiency that results in a failure to process a lipid (fat) which accumulates in the brain and other tissues. It is usually fatal by the age of two or three years, and is one of several genetic disorders found more often in populations of Ashkenazi Jewish origin.

Trinucleotide Repeat Disorders: Genetic disorders that are the result of repetitious sequences of three nucleotide bases at specific gene loci.

Turner Syndrome: A genetic disorder affecting only females, marked by the presence of one X chromosome in some or all cells, or one normal and one damaged X chromosome. The disorder is marked by short stature, delayed growth of the skeleton, shortened fourth and fifth fingers, broad chest, and heart abnormalities.

Appendix 2:

Internet and World Wide Web Resources

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Internet and World Wide Web Resources

The following appendix provides URL addresses for all WWW sites referenced in the report together with a brief description for each. In addition, we have included in a separate section two further resources: other web portals of interest; and, current GROW (Genetic Resources on the Web) sites. The web portals of interest were provided to research staff by Dr. Bob Fineman, medical consultant to the project. The GROW site list was disseminated at the recent GROW meeting, held November 30, 2000, co-sponsored by the Office of Rare Diseases (NIH) and the NHGRI.

Report Website References

BioMedNet

www.bmn.com

A resource website for biological and medical researchers. Resources include: research tools; a journal collection; web links; news and comments; books and literature; and, science job listings.

www.biomednet.com/hmsbeagle

HMS Beagle: The BioMedNet Online Magazine

Centers for Disease Control and Prevention: Genetics

www.cdc.gov/genetics

Internet site of the Office of Genetics and Disease Prevention of the CDC. The site provides current information on the impact of human genetic research and gene discoveries on disease prevention and health promotion. The site includes a weekly update of news stories, scientific literature, announcements, events, and public health perspectives on advances in human genetics.

www.cdc.gov/genetics/publications/faq.htm

"Frequently Asked Questions about Genetics and Public Health"

www.cdc.gov/genetics/resources/grevolution.html

"A Public Health Perspective."

GeneSage
www.genesage.com
GeneSage is a private for-profit company the stated mission of which is to translate the promise of genetics into solutions for health professionals and consumers. It provides users of its services with a secure place to answer sensitive questions, access genetic testing, and safely store genetic information.

www.geneletter.com
The online magazine of GeneSage

Genetics And Your Practice
http://webct.isu.edu/public/GENETICS/
Genetics & Your Practice is designed to guide physicians and other health care and social services professionals in four ways: use basic knowledge in human and medical genetics to evaluate patients who have or who are at risk for developing a genetic disorder; identify patients who could benefit from genetic services; improve access to referrals, screening, testing, diagnosis and interventions for affected or at risk individuals; and, address the financial, ethical, legal and social issues inherent in the practice of genetic medicine.

Genetics in Primary Care (GPC)
http://bhpr.hrsa.gov/dm/genpc.html
Genetics in Primary Care: A Faculty Development Initiative is funded by MCHB, the Bureau of Health Professions, NHGRI, and the Agency for Health Care Policy and Research. Its mission is to enhance the ability of faculty to incorporate genetics information into undergraduate and graduate primary care medical educations.

Human Genome Project Information: U.S. Department of Energy
www.ornl.gov/hgmis
This site, funded by the U.S. Department of Energy, introduces users to the basics about the Human Genome Project: what it is; its progress, history, and goals; frequently asked questions; and, other information for people new to the project. It also provides information on the work conducted and projects funded by the DOE ELSI Program (www.ornl.gov/hgmis/resource/elsiprog.html).

Lexis-Nexis
www.lexis-nexis.com
Lexis-Nexis is a comprehensive web-based search engine for literature and materials in a variety of media.

National Coalition for Health Professional Education in Genetics
www.nchpeg.org
The NCHPEG is a national effort to promote health professional education and access to information about advances in human genetics. NCHPEG members are an interdisciplinary group of leaders from: over 100 diverse health professional organizations; consumer and voluntary groups; government agencies; private industry; managed care organizations; and, genetics professional societies. By facilitating frequent and open communication between stakeholder groups, NCHPEG seeks to capitalize on the collective expertise and experience of members and to reduce duplication of effort.

National Human Genome Research Institute (NHGRI)
www.nhgri.nih.gov
The NHGRI's mission is to head the efforts of the National Institutes of Health (NIH) in the Human Genome Project. NHGRI is one of 24 institutes, centers, or divisions that make up the NIH.

National Human Genome Research Institute: Ethical, Legal and Social Issues (ELSI) of Human Genetics Research
www.nhgri.nih.gov/ELSI/
The ELSI Program of the NHGRI addresses the ethical, legal, and social issues that arise as the result of human genetic research. This site contains information on the program, and the research that it has funded.

National Newborn Screening and Genetics Resource Center
http://genes-r-us.uthscsa.edu
This website provides information and resources in the area of newborn screening and genetics to benefit health professionals, the public health community, consumers, and government officials.

The Pew Internet & American Life Project
www.pewinternet.org
Includes findings of a project designed to understand the importance of the Internet in health-seeking behavior.

Resource Informagen
http://www.informagen.com
Internet company that provides free directory listings for firms and businesses involved in biotechnology.

Other Internet Portals of Interest

Access Excellence
www.accessexcellence.com
Launched in 1993, AE is a national educational program that provides high school biology and life-sciences teachers access to their colleagues, scientists, and critical sources of new scientific information via the Internet.

Agency for Healthcare Research and Quality (AHRQ)
www.ahrq.gov
AHRQ supports research for improving outcomes and quality of health care, reducing health care costs, addressing medical safety, and improving access to services. This site describes AHRQ and provides research findings and health care information for consumers, providers, and researchers.

All About Genetics
http://genetics.about.com/science/genetics/mbody.htm
This website contains information about a variety of subjects including: basic genetics; biotechnology; cloning; disease and illness; education; food and agriculture; human genome; informatics; jobs; journals; mapping; news; organizations; and, research tools and testing.

Alzheimer's Research Forum
www.alzforum.org/members/index.html
The Forum was established for the purpose of enhancing information access and promoting collaboration both within the traditional Alzheimer's research community and across the numerous scientific disciplines that can contribute to the global effort to understand and treat Alzheimer's disease.

American Academy of Family Physicians
www.aafp.org
The AAFP is the national association of family doctors. The Academy was founded in 1947 to promote and maintain high quality standards for family doctors who are providing continuing comprehensive health care to the public.

American Academy of Pediatrics
www.aap.org
The AAP was founded in 1930. Members dedicate their efforts and resources to the health, safety and well-being of infants, children, adolescents and young adults.

American Cancer Society
www.cancer.org
The ACS is a nationwide, community-based, voluntary health organization dedicated to eliminating cancer as a major health problem. This website provides a broad range of scientific, research, advocacy, service, and education information about cancer.

American College of Medical Genetics
www.faseb.org/genetics/acmg
The ACMG is an organization composed of biochemical, clinical, cytogenetic, medical and molecular geneticists, genetic counselors and other health care professionals committed to the practice of medical genetics.

American College of Obstetricians and Gynecologists
www.acog.org
Founded in 1951, the ACOG is a membership organization of Obstetrician-Gynecologists dedicated to the advancement of women's health through education, advocacy, practice and research.

American College of Physicians
www.acponline.org
The mission of the ACP is to enhance the quality and effectiveness of health care by fostering excellence and professionalism in the practice of medicine.

American Diabetes Association
www.diabetes.org
The ADA is a community-based, nationwide, voluntary health organization dedicated to eliminating diabetes as a significant health problem. This website contains a broad range of scientific, research, advocacy, service and education information about diabetes.

American Heart Association
www.americanheart.org
The AHA is a nationwide, community-based, voluntary health organization dedicated to eliminating heart disease as a significant health problem. This website contains a broad range of scientific, research, advocacy, service and education information about heart disease.

American Medical Association
www.ama-assn.org
Founded in 1847, the AMA sees its members as: "much more than an organization of physicians. The AMA serves as the patient's advocate and the physicians voice." Its website was launched in 1995 with a mission to: "promote the art and science of medicine and the betterment of public health."

American Medical Association Alliance
www.ama-assn.org/alliance
The AMA Alliance is the volunteer arm of the AMA, comprised of physicians' spouses. The Alliance is dedicated to promoting better health, ensuring sound health legislation, and fund-raising for medical education.

American Public Health Association
www.apha.org
The APHA is the oldest and largest organization of public health professionals in the world. It influences public health policy and practice through professional meetings and in-service training, publications, educational services, and, advocacy efforts.

American Society of Human Genetics
www.faseb.org/genetics/ashg/ashgmenu.htm
The ASHG was established in 1948 to provide leadership in research, education and service in human genetics. Accordingly it publishes The American Journal of Human Genetics and sponsors an annual research meeting. The human genetics community grew and spawned a new field of endeavor, genetic counseling, to support delivery of clinical genetics services. Over 5,000 members include researchers, academicians, clinicians, laboratory practice professionals, genetic counselors, nurses and others involved in human genetics.

Biology Website References for Students and Teachers
www.hoflink.com/~house/MendelGen.html
www.hoflink.com/~house/MolecularGen.html
These two reference websites contain a multitude of hot links for students and teachers regarding basic genetics, and human and medical genetics, including: molecular genetics; gene mapping; chromosomes and karyotypes; inheritance; cloning; and, the genetics of specific conditions.

Biotechnology Industry Organization
www.bio.org/welcome.html
Website for professionals in the biotechnology industry. The website includes information on: membership; government affairs; worldwide biotech events; news, services for members, current topics and links to related organizations.

California State Department of Health Services
www.dhs.ca.gov/applications/search/Search.asp
A search for the term "genetics" at this state department of health website provides an example of how a state government is planning for and addressing issues related genetics in public health.

Celera Genomics
www.celera.com
The Celera Discovery System provides integrated sequence data and analysis tools to the not-for-profit, government, and academic research community.

Center for Inherited Disease Research
www.cidr.jhmi.edu
CIDR is a centralized facility established to provide genotyping and statistical genetics services for investigators seeking to identify genes that contribute to human disease. CIDR concentrates primarily on multifactorial hereditary diseases, although linkage analysis of single gene disorders can also be accommodated.

Child Development Center: National Center for Cultural Competency
http://gucdc.georgetown.edu/cultural.html
The mission of the NCCC is to increase the capacity of health care programs to design, implement and evaluate culturally competent service delivery systems.

Coalition of State Genetics Coordinators
www.stategeneticscoordinators.org
The Coalition is an organization of state and territorial genetics coordinators. The mission of the Coalition is to promote core public health functions as they apply to genetics.

Communities of Color and Genetics Policy Project
www.sph.umich.edu/genpolicy
Michigan State University Center for Ethics and Humanities in the Life Sciences and Tuskegee University National Center for Bioethics in Research and Health Care have combined efforts to form a five year project designed to provide policy recommendations based on public perceptions and responses to the explosion of genetic information and technology. The project also tests the process of community dialogue as an effective means to engage citizens in thoughtful and productive discussions about policy needs regarding genetic information and technology.

Continuing Medical Education Credits
www.cmesearch.com
CMESearch.com is one of the largest continuing medical education (CME) sites on the Internet offering the most comprehensive free list of accredited resources for physicians including detailed information regarding program contents, speakers/faculty, accreditation, and any special offerings.

Council for Responsible Genetics
www.gene-watch.org/org.html
The CGR is a national organization of scientists, environmentalists, public health advocates, physicians, lawyers and other concerned citizens. CRG encourages informed public debate about the social, ethical, and environmental implications of new genetic technologies, and advocates for socially responsible use of these technologies. It also monitors the development of new genetic technologies in two broad program areas: human genetics; and, commercial biotechnology and the environment.

Criteria for Assessing the Quality of Health Information on the Internet
http://hitiweb.mitretek.org/iq
Mitretek Systems supports The Health Summit Working Group's "Criteria for Assessing the Quality of Health Information on the Internet." It includes a tool to assist consumers in learning how to ask the right questions and evaluate the strengths and weaknesses of websites.

Dictionary of Cell Biology
www.mblab.gla.ac.uk/~julian/dict.html
A searchable website dictionary of words and terms relevant to cell biology and genetics.

DNA Learning Center
http://vector.cshl.org
A science center devoted entirely to public genetics education. Special emphasis has been placed on teaching genetics at the pre-college level. This center is an operating unit of the Cold Spring Harbor laboratory.

drkoop.com
www.drkoop.com
A health resource website. A wide variety of topics are covered and information is easily accessible.

Economics and Net Medical Ethics
www.msnbc.com/news/420584.asp#BODY
Editorial article written by msnbc columnist Glenn McGee, Ph.D.

Education Appraisal Skills
www.appraisalskills.com
This educational website was developed to provide medical trainers and trainees in hospital medicine and in general practice with the opportunity to use web-based technology to help develop skills in educational planning and appraisal.

Educational Development Center
www.edc.org/
The website for a non-profit education and health organization that brings researchers and practitioners together to create tools and conditions for learning, reaching people of all ages, backgrounds and abilities.

elearningpost
www.elearningpost.com
elearningpost's mission is to provide quality e-learning content that attracts a diverse and emerging audience through daily links, featured articles, and related special reports.

Facing Our Risk of Cancer Empowered (FORCE)
www.facingourrisk.org
The goals of FORCE are to: "provide support for women at high risk of breast and ovarian cancer due to genetic predisposition, family history or other factors; provide support for families facing these risks; provide women with resources to determine if they are at high risk for hereditary breast and ovarian cancer; and raise funds for research in the area of hereditary breast and ovarian cancer."

Family Village
www.familyvillage.wisc.edu/index.htmlx
The Family Village is a global community that integrates information, resources, and communication opportunities on the Internet for persons with cognitive and other disabilities, for their families, and for those that provide them services and support. The community includes informational resources on specific diagnoses, communication connections, adaptive products and technology, adaptive recreational activities, education, worship, health issues, disability-related media and literature.

FATHOM
www.fathom.com
A site for interactive knowledge and online education with specific "Health and Medicine" sections.

Fetal Diagnosis
www.TheFetus.net
This site reviews, on a regular basis, different case reports involving fetal conditions and prenatal diagnostic issues.

Foundation for Blood Research
www.fbr.org
The FBR finds more effective ways to identify, manage and prevent human disease through: clinical and laboratory investigation; epidemiology; outreach

science education; computer-based analysis; public health program design; population screening; and, clinical testing.

Foundation for Genetic Education and Counseling
www.fgec.org
The Foundation was established to promote understanding of human genetics and genetic medicine among health professionals and the public. The aim of this site is to provide informative, current and engaging views of the nature and implications of research in human genetics.

Foundation for Genetic Medicine, Inc.
www.GeneticMedicine.org
The FGM has two prime objectives: 1) To examine critical issues and help develop and gain acceptance for public policies which support and advance genetic medicine for improved human health; and, 2) To help society achieve "genomic literacy" through education about genetic medicine and research, and their ethical, legal and social dimensions and implications.

GEMdatabase
www.dnai.com/~pboyd/CORN/worksinp.htm
The GEM (Genetic Education Materials) database lists and describes publications produced by federally funded regional genetics networks and other federally funded projects. Links to ordering information and, when available, the on-line version of each publication are provided.

GeneClinics
www.geneclinics.org
A clinical information resource relating genetic testing to the diagnosis, management, and genetic counseling of individuals and families with specific inherited disorders.

Geneforum
www.geneforum.org
Geneforum helps users of this website understand recent advances in genetics and technology, and to address important/related ethical, legal, and social questions and issues.

GeneLetter
www.geneletter.com
An online magazine (published by GeneSage) of genetics, society and culture.

Gene Med Network
www.genemed.org
The mission of Gene Med Network is to provide reliable, up-to-date news and information on gene therapy and to introduce other gene therapy related resource websites for health care professionals, basic scientists, people working in biotechnology based industries, consumers, and the general public. The site includes: news updates; general information on gene therapy; journal articles; consulting reports;and, links to related sites.

Genes at Work: Center of Human and Molecular Genetics
www.genesatwork.org
Genes at Work's goal is to provide primary care providers with the required tools to address the genetic needs of their patients. The site includes access to specialty-specific information and foreign language educational materials for patients.

GeneTests
www.genetests.org
Funded by the National Library of Medicine of the NIH and Maternal and Child Health Bureau of HRSA, GeneTests is a genetic testing resource that includes: a genetics laboratory directory; a genetics clinic directory; an introduction to genetic counseling and testing concepts (in, "About Genetic Services"); a PowerPoint slideshow presentation for genetics professionals (in, "Teaching Tools"); and, a link to disease-specific profiles in the companion project, GeneClinics.

Genetic Alliance
www.geneticalliance.org
The Genetic Alliance is an international coalition of individuals, professionals and genetic support organizations that is working together to enhance the lives of everyone impacted by genetic conditions.

Genetic Education Database
http://genetics-education.mbt.washington.edu/database
This site includes a comprehensive directory of programs, resources and information for students and teachers. This site is maintained by the High School Human Genome Program at the University of Washington.

Genetic Health
www.geneticheath.com
Genetic Health enables consumers to learn the latest about genetics and their health, to assess their inherited risk on line, and act to take control of their own health.

Genetic Interest Group
www.gig.org.uk
A British National Alliance of organizations that support children, families and individuals affected by genetic disorders. Its primary goal is to promote awareness and understanding of genetic disorders so that higher quality services for people affected by genetic conditions are developed and made available to all who need them.

Genetics & Ethics
www.ethics.ubc.ca/brynw/
This website describes a wide variety of ethical issues in genetics. It contains the names and locations of ethicists interested in genetics, references, and information on specific topics like gene therapy, testing, privacy, and discrimination.

Genetics & Molecular Medicine Front Page: American Medical Association
www.ama-assn.org/ama/pub/category/1799.html
This website provides education, advocacy and news to physicians, health care providers and consumers.

Genetics for Health Professionals
www.nurs.uic.edu/genetics
This website aims to educate health care professionals about the importance of genetics in their practice. It includes: lectures about molecular genetics; screening and testing; specific genetic conditions; ethical, legal and social issues; case reports; and, reference and resource information.

Genetics in Medicine
www.wwilkins.com/GIM
The official journal of the American College of Medical Genetics. Its mission is to enhance the knowledge and practice of medical genetics.

Genetics of Cancer
www.cancergenetics.org
The purpose of this website is to provide information that will help users understand the genetic basis of cancer, and to help users interpret new discoveries in the field of cancer genetics.

Genetics Program for Nursing Faculty: Children's Hospital of Cincinnati
www.gpnf.org
GPNF is a multi-faceted genetics educational program for nursing faculty. GPNF's goals are to increase nursing faculty knowledge about human genetics

and its clinical applications and to increase the amount of human genetics content and clinical experiences taught in RN preparatory educational programs.

Genetics Resource Center
www.pitt.edu/~edugene/resource
The Genetics Resource Center is an online resource and starting point for genetics counseling related information. The website is constructed and maintained by the Genetics Education and Counseling Program at the University of Pittsburgh.

Genetics Virtual Library
www.ornl.gov/TechResources/Human_Genome/genetics.html
This website is a subject catalog covering genetics in the biosciences.

Genome Action Coalition
www.tgac.org
The GAC is comprised of patient advocacy organizations, professional organizations in the field of genetics and genomics, consumer organizations, university-based research facilities, pharmaceutical research companies and biotechnology companies. The Coalition exists to promote an environment in government and in the private sector in which genome research can continue to flourish.

Hardin Meta Directory of Internet Health Sources
www.lib.uiowa.edu/hardin/md
The Hardin Meta Directory lists sources for health-related websites.

Hastings Center
www.thehastingscenter.org
The Hastings Center is an independent, nonpartisan, interdisciplinary research institute that addresses fundamental ethical issues in the areas of health, medicine and the environment as they affect individuals, communities and societies.

Healthfinder
www.healthfinder.gov
This website is a service of the U.S. Department of Health and Human Services which provides a free guide to reliable health information.

Health Internet Ethics
www.hiethics.org
This site unites the most widely used health Internet sites supporting the "highest ethical standards." Their goal is to establish and comply with the

highest standards for privacy, security, credibility and reliability so that consumers can realize the fullest benefits of the Internet.

Health Legacy Partnership (HELP)
www.healthlegacy.org
The mission of the Health Legacy Partnership, a partnership between The Kanter Family Foundation and the Agency for Healthcare Research and Quality (AHRQ), is to establish a database of health outcomes information on a wide range of disorders and conditions for consumers and providers.

Howard Hughes Medical Institute
www.hhmi.org/genetictrail/
This website contains a report written in 1997 from the Howard Hughes Medical Institute entitled, "Blazing a Genetic Trail: Families and Scientists Join in Seeking the Flawed Genes that Cause Disease."

Human Genome Education Model Project: Part II
www.georgetown.edu/research/hugem
The purpose of the HuGem II Project is to provide educational training and resources for members of seven collaborating professional organizations to increase the knowledge and sensitivity to human genetics, the Human Genome Project, and the ethical, legal, and psychosocial issues of genetic testing and research.

Human Genome Epidemiology Network
www.cdc.gov/genetics/hugenet/default.htm
HuGE Net is a global collaboration of individuals and organizations committed to the development and dissemination of population-based epidemiologic information on the human genome. The site features information on: population-specific prevalence data on human gene variants; epidemiologic data on the association between genetic variation and diseases in different populations; quantitative population-based data on gene-environment interaction; and, population impact on the use of genetic tests and services in improving health and preventing disease.

Human Genome Project: Introduction
www.accessexcellence.org/AB/IE/Intro_The_Human_Genome.html
An introduction to and description of the Human Genome Project including ethical issues and a graphics gallery.

Human Molecular Genetics
www.hmg.oupjournals.org/
An on-line journal concentrating on full-length research papers covering a wide range of topics in all aspects of human molecular genetics. A free, e-mail table of contents alerting service is also available.

INFOGENETICS
www.infogenetics.org
A comprehensive genetics database. In addition, information on directories of support groups and clinical care guidelines are included.

Institute for Clinical Evaluation
www.icemed.org
The ICE is a nonprofit educational organization whose purpose is to improve the quality of health care available to the public. ICE accomplishes its mission by: creating and administering high-quality and valid testing processes for health care professionals; setting clinical performance standards; identifying clinicians who meet these standards and offering credentials that attest to their competence; and, informing consumers about the skill level of their health care providers.

Institute for Genomic Research
www.tigr.org/
A research institute with interests in structural, functional, and comparative analysis of genomes and gene products in viruses, eubacteria, pathogenic bacteria, archaea, and eukaryotes (both plant and animal), including humans. Located in Rockville, Maryland, TIGR has a large DNA sequencing laboratory and has modern facilities for bio-informatics, biochemistry and molecular biology.

International Communication Forum in Human Molecular Genetics
www.hum-molgen.de/
This service provides the opportunity to communicate with scientists, physicians and other genetics professionals worldwide.

International Society of Nurses in Genetics
http://nursing.creighton.edu/isong
ISONG is a nursing specialty organization dedicated to fostering the scientific and professional growth of nurses in human genetics.

Journal of Medical Internet Research
www.jmir.org
This journal is an international scientific peer-reviewed journal on all aspects of research, information and communication in the health care field using Internet and intranet-related technologies.

Kansas University Medical Center Genetic Education Project for Educators

www.kumc.edu/gec

An informative website for educators interested in human genetics and the Human Genome Project.

Kansas University Medical Center Genetic Education Project for Professionals

www.kumc.edu/gec/geneinfo.html

Information for genetic professionals at the University of Kansas Medical Center is updated regularly with clinical, research, and educational resources for genetic counselors, clinical geneticists, and medical geneticists. This is a useful resource for teachers as well.

March of Dimes: Resource Center

www.modimes.org

The Resource Center provides accurate, timely information and referral services to the public. The staff at the Resource Center includes trained professionals who help people, one on one, to address personal and complex problems. They answer questions from parents, health care providers, students, librarians, government agencies, health departments, social workers, and other people from all walks of life and from around the world.

Massachusetts State Department of Public Health

www.state.ma.us/dph/

A keyword search for "genetics" at this website provides an example of how state government can deal with the issue of genetics in public health.

Maternal and Child Health Bureau, Health Resources and Services Administration, Department of Health and Human Services

www.mchb.hrsa.gov/

The MCHB provides its leadership, partnership and resources to advance the health of all our nation's mothers, infants, children and adolescents including families with low income levels, those with diverse racial and ethnic heritages and those living in rural or isolated areas without access to care.

Maternal and Child Health (MCH) Neighborhood

http://mchneighborhood.ichp.edu/

Support and training for the development of websites for projects funded by the Maternal and Child Health Bureau. MCH Neighborhood will host your site free of charge and will provide you with exclusive password-protected remote access privileges so that you may keep your site up-to-date and dynamic from wherever you may be located (see especially /wagenetics, /pacnorgg, and /geneticlink sections).

Medline Plus Health Information
www.medlineplus.gov
A health resource website. Information covered includes health topics, drug information, dictionaries, and directories. This site is a service of the National Library of Medicine.

Medscape
www.medscape.com
Medscape's goals are: " to provide clinicians and other health care professionals with the most timely source of clinical information that is highly relevant to their patients and practice, to make the clinician's information gathering more fruitful and less time consuming, and to make available to a broad medical audience clinical information with the depth, breadth and validity needed to improve the practice of medicine."

Mid-Atlantic Regional Human Genetics Network
www.pitt.edu/~marhgn/
Serving: Delaware, Maryland, New Jersey, Pennsylvania, Virginia, Washington D.C., and West Virginia.

Mountain States Regional Genetic Services Network
www.mostgene.org
Serving: Montana, Wyoming, Utah, Colorado, Arizona, and New Mexico.

National Cancer Institute
http://cancernet.nci.nih.gov
CancerNet is the gateway to the most recent and accurate cancer information from the National Cancer Institute (NCI), a component of the NIH.

National Center for Biotechnology Information
www.ncbi.nlm.nih.gov
Established in 1988, the NCBI has been given the responsibility to: create systems for the storage of information; perform research; facilitate the use of databases (such as PubMed) and software for researchers; and, coordinate efforts to gather biotechnology information worldwide.

National Center for Genome Resources
www.ncgr.org
The staff of NCGR works in the field of bio-informatics with a goal of applying genetic data for humanity's best possible advantage.

National Heart, Blood, and Lung Institute
www.nhlbi.nih.gov/
The NHLBI provides leadership for a national program in: diseases of the heart, blood vessels, lungs, and blood; sleep disorders; and, blood resources.

National Institutes of Health
www.nih.gov
Website for the National Institutes of Health (NIH), U.S. Department of Health and Human Services.

National Institutes of Health Office of Rare Diseases
http://rarediseases.info.nih.gov/ord/
The ORD provides information on more than 6000 rare diseases, including current research, publications from scientific and medical journals, completed research, ongoing studies, and consumer support groups.

National Library of Medicine
www.nlm.nih.gov/
The website for "the world's largest medical library." In addition to the library database, this site also contains health information, research programs, news, and general information.

National Organization for Rare Disorders
www.rarediseases.org/
NORD is a unique federation of voluntary health organizations dedicated to helping people with rare "orphan" diseases and assisting the organizations that serve them. NORD is committed to the identification, treatment, and cure of rare disorders through programs of education, advocacy, research, and service.

National Parent to Parent Support & Information System, Inc.
www.NPPSIS.org
NPPSIS is a National Resource for providing emotional and informational support for parents who have children with special health care needs and/or rare disorders.

National Partnership for Women and Families
www.nationalpartnership.org
The NPWF is a nonprofit, nonpartisan organization that uses public education and advocacy to promote fairness in the workplace, quality health care, and policies that help women and men meet the dual demands of work and family.

National Society of Genetic Counselors
www.nsgc.org/
The NSGC is an advocacy organization for the genetic-counseling profession that, among other things, offers consumers referrals to genetic counselors.

New England Regional Genetics Group
www.acadia.net/nergg/index.html
Serving: Maine, Vermont, New Hampshire, Massachusetts, Rhode Island, and Connecticut.

New Genetics: A Resource for Teachers and Students
http://www4.umdnj.edu/camlbweb/teachgen.html
A comprehensive list of genetic education websites aimed at high school and college students and teachers.

New York Online Access to Health
www.noah-health.org
NOAH seeks to provide high quality full-text health information for the under-served that is accurate, timely, relevant and unbiased. NOAH supports English and Spanish.

Onhealth: In Association with WebMD
www.onhealth.com
A health resource website with a variety of topics covered in an easy to read format.

Online Mendelian Inheritance in Man
www3.ncbi.nlm.nih.gov/Omim/
This database is a catalog of human genes and genetic disorders authored and edited by Dr. Victor McKusick and his colleagues at Johns Hopkins and elsewhere, and developed for the Internet by NCBI, the National Center for Biotechnology Information. The database contains textual information, pictures, and reference information. It also contains numerous links to NCBI's Entrez database of MEDLINE articles and sequence information.

Pacific Northwest Regional Genetics Group
http://mchneighborhood.ichp.edu/pacnorgg
Serving: Alaska, Idaho, Oregon, and Washington.

Pacific Southwest Regional Genetics Network
www.psrgn.org
Serving: California, Hawaii, and Nevada.

Pew Charitable Trusts: Health and Human Services
www.pewtrusts.com
This agency designed their Health and Human Services program to promote the health and well-being of the American people and to strengthen disadvantaged communities. Currently, Pew offers national and local grants in the areas of public health and health/biomedical research.

Pharmaceutical Research & Manufacturers of America: Genomics
www.phrma.org/genomics/
A comprehensive resource site about genomics and related information.

Praxis.md: Practical Answers for Patients and Physicians
www.praxis.md
A reference website for patients and physicians. Information is comprehensive and continually updated.

Public Health Genetics Society: University of Michigan
www.umich.edu/~phgs/
The goal of the Public Health Genetics Society is to promote awareness of the role of genetics in public health and disease and the implications of genetic technology on public health. The Society hopes to fulfill this goal by publishing a newsletter and sponsoring presentations, seminars, and conferences related to these issues.

Rare Genetic Diseases in Children
http://mcrcr2.med.nyu.edu/murphp01/homenew.htm
An Internet jump-station to sources of information on rare genetic diseases affecting children. Under the aegis of the NYU Medical Center, this site has provided its services since 1996.

Robert Wood Johnson Foundation
www.rwjf.org/
The RWJF's mission is to improve the health and health care of all Americans, from encouraging healthier living and the conditions that promote better health, to promoting positive changes in the way health care is delivered in this country. Their priority areas are access to care, substance abuse, and chronic care.

Science and Technology: PBS
www.pbs.org/search/
Users may search the PBS website for "genetics" related issues. More than 1,000 matches can be found relating to a wide variety of fundamental and clinically-related issues.

Science Panel on Interactive Communication and Health
www.scipich.org/pubs/pubs.htm
This site contains a series of journal articles and a final report intended to accelerate the appropriate development, adaptation, use, and evaluation of interactive health communication applications.

Search the Virtual Hospital
www.vh.org/Misc/Search.html
This website enables you to search the contents of all of the documents in the University of Iowa Medical Center's Virtual Hospital (see especially, Clinical Genetics: A Self Study for Health Care Professionals at www.vh.org/Providers/Textbooks/ClinicalGenetics/Lession1/L1Contents.html).

Secretary's Advisory Committee on Genetic Testing (Department of Health and Human Services)
www4.od.nih.gov/oba/sacgt.htm
Donna Shalala, Secretary of Health and Human Services, chartered the SACGT in 1998 to write and disseminate the report entitled, "Enhancing the Oversight of Genetic Tests."

Sickle Cell Disease Information Center
www.emory.edu/PEDS/SICKLE
The mission of this site is to provide consumers with sickle cell disease and health care professionals with education, news, research updates, and world wide sickle cell resources. It is the mission of their organizations to provide compassionate care, education, counseling, and research for patients with sickle cell disease.

Southeastern Regional Genetics Group
www.cc.emory.edu/PEDIATRICS/sergg/
Serving: Alabama, Florida Georgia, Kentucky, Louisiana, Mississippi, North Carolina, South Carolina, and Tennessee.

Special-Needs Collection: Woodbine House Publishers
www.woodbinehouse.com
Woodbine House publishes *The Special-Needs Collection*, a series of about 50 books for parents and health and social services professionals regarding disabilities and related topics, e.g., ADD, autism, Down's Syndrome, dyslexia, Fragile-X, spina bifida.

Task Force on Genetic Testing Report: March, 1996
http://infonet.welch.jhu.edu/policy/genetics/intro.html
The TFGT was convened to review genetic testing in the U.S. and make recommendations to ensure the development of safe and effective genetic tests,

their delivery in laboratories of assured quality, and their appropriate use by health care providers and consumers.

Technology Museum of Innovation
www.thetech.org/exhibits_events/online/genome/overview.html
The purpose of The Tech is to help educate, enlighten and inspire the next generation of innovators, i.e., students grades K-12.

Teratology Society
www.teratology.org
This website provides information about childhood development and birth defects at the fundamental and clinical levels, as well as appropriate means of prevention.

Texas Genetics NetworK
www.tdh.state.tx.us/texgene/texgene.htm
Serving: Texas and the surrounding states

TRUSTe: Building a Web You Can Believe In
www.truste.org/
This website is for web publishers as well as web users. It provides information and resources regarding the protection of your privacy while using the Internet.

U.S. Congress on the Internet: Library of Congress
http://thomas.loc.gov/home/thomas.html
Access to the latest congressional information by legislative bill or a particular issue is provided on this website this is an excellent website (for example, search on the word "genetics").

U.S. Pharmacopeia
www.usp.org
USP helps to ensure that consumers receive medicines of the highest possible quality by setting the standards that manufacturers must meet to sell their products in the U.S. As the "world's most highly recognized and technologically advanced Pharmacopeia," USP provides standards for more than 3,700 medications, dietary supplements and dosage forms.

WebMD
www.webmd.com
A health resource site with specific information for: physicians, consumers, office managers and health teachers.

World of Genetic Societies
www.faseb.org/genetics
This site serves as a directory of genetics professional and other related sites.

Current Grow (Genetics Resources on the Web) Sites

The sites listed below reflect the current membership of the GROW organization and their websites as of 11/30/2000. The GROW search engine is temporarily located at http://search.info.nih.gov/grow

American Association for the Advancement of Science (AAAS)
www.aaas.org/

American College of Medical Genetics
www.faseb.org/genetics/acmg

American Medical Association
www.ama-assn.org

American Society of Human Genetics
www.ashg.org

Celera Genomics
www.celera.com

Centers for Disease Control and Prevention
www.cdc.gov/genetics/

Children's Hospital of Cincinnati
www.gpnf.org

Department of Energy
www.ornl.gov/ghmis/resource/assist.html

DNA Dynamics, Inc.
www.dnaMD.com

Education Development Center
www.edc.org

Foundation for Genetic Education and Counseling
www.fgec.org

Foundation for the National Institutes of Health
www.fnih.org

GeneClinics
www.geneclinics.org

GeneSage
www.genesage.com

GeneTests
www.genetests.org

The Genetic Alliance
www.geneticalliance.org

Genetic Health
www.geneticheath.com

Genomic Health
www.genomichealth.com

Genetics in Medicine
www.wwilkins.com/GIM

Global TeleGenetics, Inc
www.GeneScene.com

INFOGENETICS
www.infogenetics.org

International Society of Nurses in Genetics
www.nursing.creighton.edu/isong

Kansas University Medical Center
www.kumc.edu/gec/geneinfo.html

March of Dimes Birth Defects Foundation
www.modimes.org

Maternal and Child Health Bureau (HRSA)
www.mchb.hrsa.gov/genetics.htm

National Cancer Institute
www.cancernet.nci.nih.gov

National Center for Biotechnology Information
www.ncbi.nlm.nih.gov

National Coalition for Health Professional Education in Genetics
www.nchpeg.org

National Human Genome Research Institute
www.nhgri.nih.gov

National Institutes of Health – Office of the Director
www.nih.gov

National Institutes of Health – Office of Extramural Research
www.nih.gov/sigs/bioethics

National Organization for Rare Disorders
www.rarediseases.org/

National Society of Genetic Counselors
www.nsgc.org

Nature
www.nature.com/genomics

Office of Rare Diseases, NIH
www.rarediseases.info.nih.gov/ord/

Orchid BioSciences, Inc.
www.geneshield.com

Parmaceutical Research & Manufacturers of America
www.phrma.org/genomics

U.S. Pharmacopeia
www.usp.org