# Table of Contents

Acknowledgements...........................................................................................................ii  
Definitions.........................................................................................................................iii  
Glossary...............................................................................................................................iv  
Executive Summary...........................................................................................................v  
Chapter 1: Background......................................................................................................1  
Chapter 2: Illinois State Profile........................................................................................4  
Chapter 3: Current Status...................................................................................................29  
Chapter 4: Survey Findings: Local Health Departments...................................................53  
Chapter 5: Survey Findings: Genetic Service Providers....................................................66  
Chapter 6: Interview Findings............................................................................................84  
  Genetic Service Providers.................................................................................................84  
  Non-Genetic Health Care Providers..............................................................................105  
  Local Health Department Personnel............................................................................108  
  State Agency Personnel.................................................................................................111  
  Consumer/Advocacy Group Representatives.................................................................114  
Chapter 7: Public Attitudes and Perceptions....................................................................117  
Chapter 8: State Laws and Legislation..............................................................................128  
Chapter 9: Discussion/Next Steps....................................................................................134  

Appendices:  
  1: Methods....................................................................................................................149  
  2: LHD Survey Questionnaire.........................................................................................155  
  3: Genetic Services Provider Survey Questionnaire.......................................................161  
  4: Interview Guides........................................................................................................172  
  5: List of Disorders Screened..........................................................................................182  
  6: List of IDPH Local Health Department Grantees.......................................................183  
  7: IDPH Expanded Screening Tool...............................................................................186  
  8: Genetics and Metabolic Disease Advisory Committee  
      (GMDAC) Members..................................................................................................189  
  9: Catalogue of On-Going Medical Genetics Projects in Illinois.................................191
  by Institution
Acknowledgements

We wish to thank all those who contributed to this study and final report. We would especially like to thank:

- Claudia Nash, State Genetics Coordinator, Luna Okada, Project Coordinator, and Karen Burget, Nurse Consultant, Genetics and Newborn Screening Program, Division of Health Assessment and Screening, Illinois Department of Public Health, for their support and input throughout this project.

- Hollis Russinof, for her contributions to the writing of this report.

- Staff from the Midwest Center for Health Workforce Studies, University of Illinois at Chicago: Louise Martinez and Rocio Ruiz, for their expertise and efforts in formatting and editing this report; and Antonia Sweet, for her research assistance.

- Judith Cooksey, University of Maryland at Baltimore, for bringing this project to us, assisting in the development of the project proposal, and for her thoughtful questioning and support throughout the project.

- Judith Benkendorf and Helen Travers, for assisting in the development of the project proposal.

- Janice Bach, Roslyn Beene, Kerry Silvey, and Debra Doyle, for helping us to conceptualize this project.

Finally, we would like to acknowledge all of the survey respondents and interview participants. Your time and input was invaluable to us during this project, and will continue to prove so as we develop a plan for genetic services in Illinois.

This project was funded by the Illinois Department of Public Health.
Definitions

Genetic counselors\(^1\) Genetic counselors are health professionals with specialized graduate degrees and experience in the areas of medical genetics and counseling.

Genetic disease\(^2\) Sickness, physical disability, or other disorder resulting from the inheritance of one or more deleterious forms of a gene.

Genetic screening\(^3\) Testing a group of people to identify individuals at high risk of having or passing on a specific genetic disorder.

Genetic testing\(^2\) Analyzing an individual’s genetic material to determine predisposition to a particular health condition or to confirm a diagnosis of genetic disease.

Genetics\(^4\) The science of heredity; the study of genes and the way they determine traits and characteristics passed from generation to generation. In contrast to genomics, “genetics” refers to a single gene and its effects.

Genome\(^3\) All the DNA contained in an organism or a cell, which includes both the chromosomes within the nucleus and the DNA in the mitochondria.

Genomic Competencies\(^5\) The minimum knowledge, skills, and attitudes necessary for health professionals from all disciplines to provide patient care that incorporates genetic perspectives and reflects sensitivity to related ethical, legal, and social concerns.

Genomics\(^4\) The study of the entire genome, including the complex interactions among multiple genes as well as between genes and the environment. Applied to public health, genomics offers the potential to better understand the roles of genes, environment, and behavior as risk factors for complex chronic diseases.

Human Genome Project\(^3\) An international research project to map each human gene and to completely sequence human DNA.

Medical geneticist\(^6\) A medical doctor or medical researcher who has completed a fellowship or has other advanced training in medical genetics.

Needs assessment\(^2\) A method or process used to determine if a need exists for a given program or service. A need can be defined as a discrepancy between what “is” and what should be.

Newborn screening\(^4\) A public health program mandated by state law to test newborns for certain rare but treatable disorders.

## Abbreviations

<table>
<thead>
<tr>
<th>Abbreviation</th>
<th>Description</th>
</tr>
</thead>
<tbody>
<tr>
<td>ABGC</td>
<td>American Board of Genetic Counseling</td>
</tr>
<tr>
<td>ABMG</td>
<td>American Board of Medical Genetics</td>
</tr>
<tr>
<td>ABMS</td>
<td>American Board of Medical Specialties</td>
</tr>
<tr>
<td>APORS</td>
<td>Adverse Pregnancy Outcomes Reporting System</td>
</tr>
<tr>
<td>ASHG</td>
<td>American Society of Human Genetics</td>
</tr>
<tr>
<td>ASTHO</td>
<td>Association of State and Territorial Health Directors</td>
</tr>
<tr>
<td>CDC</td>
<td>Centers for Disease Control and Prevention</td>
</tr>
<tr>
<td>CSHCN</td>
<td>Children with Special Health Care Needs</td>
</tr>
<tr>
<td>DSCC</td>
<td>Division of Specialized Care for Children</td>
</tr>
<tr>
<td>ELSI</td>
<td>Ethical, Legal, and Social Issues</td>
</tr>
<tr>
<td>GIPA</td>
<td>Genetic Information Privacy Act</td>
</tr>
<tr>
<td>GME</td>
<td>Graduate Medical Education</td>
</tr>
<tr>
<td>GTTFI</td>
<td>Genetic Task Force of Illinois</td>
</tr>
<tr>
<td>HB</td>
<td>House Bill</td>
</tr>
<tr>
<td>HRSA</td>
<td>Health Resources and Services Administration</td>
</tr>
<tr>
<td>IDHS</td>
<td>Illinois Department of Human Services</td>
</tr>
<tr>
<td>IDPH</td>
<td>Illinois Department of Public Health</td>
</tr>
<tr>
<td>ISONG</td>
<td>International Society of Nurses in Genetics</td>
</tr>
<tr>
<td>NCHPEG</td>
<td>National Coalition for Health Professional Education in Genetics</td>
</tr>
<tr>
<td>NHGRI</td>
<td>National Human Genome Research Institute</td>
</tr>
<tr>
<td>NSGC</td>
<td>National Society of Genetic Counselors</td>
</tr>
<tr>
<td>PA</td>
<td>Public Act</td>
</tr>
<tr>
<td>PCP</td>
<td>Primary care provider</td>
</tr>
<tr>
<td>SB</td>
<td>Senate Bill</td>
</tr>
</tbody>
</table>
Executive Summary

The Centers for Disease Control and Prevention has articulated a vision of public health in the 21st century that emphasizes the integration of genetics/genomics into public health practice. As a part of this vision, the CDC has encouraged every state to develop a genetics plan identifying the specific ways in which states will address their current and future genetic service and policy needs. Planning in this arena is crucial at this time because of the rapid development of genetic science and technology, the actual and potential consequent changes in the medical care delivery system, the impact the field will have on population health, and associated broad social and policy implications.

The first step in the process of developing a state plan is the completion of a needs assessment. The Illinois Department of Public Health contracted with the Midwest Center for Health Workforce Studies, located in the School of Public Health at the University of Illinois at Chicago to conduct a statewide genetics needs assessment.

The purpose of this needs assessment is to assess the role and function of the state health department with regard to the current and future needs for genetic policy, service provision, workforce and community education, as well as research and surveillance in Illinois. This was accomplished through:

- An assessment of the current provision of genetic services in Illinois; and,
- An assessment of the essential elements of a state genetics program, currently and in the coming years, defined through the perspectives of key stakeholders in Illinois, and the visions and practices of key state and national public health agencies.

The needs assessment process involved the review of key documents and program literature, as well as surveys and interviews conducted with key stakeholders throughout the state. Surveys were sent to genetic coordinators or maternal and child health coordinators at all 94 local health departments and to all 150 identified genetic service providers in Illinois, including medical and laboratory geneticists, genetic counselors, and genetic nurses. Responses were received from 71 health departments and 78 genetic service providers. A total of 86 interviews were conducted with genetic service providers, other health care providers, local health department personnel, state agency staff, and representatives of advocacy groups.

This report provides an overview of Illinois’ population characteristics, summarizes the current genetic services system in Illinois, presents findings from the surveys and interviews, reviews literature about public attitudes and perspectives of ethical, legal, and social issues (ELSI) in genetics, and summarizes key legislation in Illinois relevant to genetic services. It includes the information that will be used to develop goals and objectives, and priorities, for a genetic services state plan for Illinois.

Overview of Current System

The needs assessment includes descriptive data about Illinois’ population sociodemographics, health insurance, and health status, as well as the current genetic services system. The Illinois Department of Public Health’s (IDPH) Genetics and Newborn Screening Program is the key coordinator of public health genetics in the state. The program has the following responsibilities: administering the state’s legally mandated universal newborn
screening program: coordinating and overseeing a statewide public/private network of clinical
genetic centers and local health departments; and providing educational programs to the public
and health care providers on genetics and health promotion. In addition to IDPH, the Illinois
Department of Human Services (IDHS), the administrator of the Title V Maternal and Child
Health Block Grant, and the Division of Specialized Care for Children (DSCC), the state’s Title
V Children with Special Health Care Needs Agency, provide case management, clinical care,
and support services to women and children with reproductive and special health care needs,
including those associated with certain types of genetic conditions and/or indications associated
with genetic components or risk factors.

Key Findings

Below are summaries of the key findings from: the surveys of genetic service providers
and local health departments; the interviews with genetic service providers, non-genetic health
care providers, local health department personnel, state agency personnel, and
consumer/advocacy group representatives; a review of literature regarding public attitudes and
perceptions of ethical, legal and social issues (ELSI) in genetics; and a review of Illinois state
laws and legislation.

Genetic Service Providers

Survey Results. This section provided demographic and practice data for genetic service
providers in Illinois. It is important to note that the results from these survey respondents may
not be generalizable to all genetic providers in Illinois, although the results are consistent with
the recently published results of a national survey.

Almost all genetic providers were white and non-Hispanic. Over half of geneticists and
all but two genetic counselors were women. The youngest geneticist respondent was 41 years of
age, with 34% aged 55 years or older. In contrast, 60% of genetic counselors were less than 35
years of age, and almost half (47%) had been at their current place of employment for less than
five years. On average, geneticists worked 51 hours per week, while genetic counselors worked
40 hours. Only 8% reported that their practice was full.

Eighty percent of respondents worked either in an academic medical center/university or
a hospital. About half of respondents were comfortable with staffing levels at their primary work
setting. Most providers discussed risks with patients when recommending susceptibility testing
or pre-symptomatic testing. There was support for potential telemedicine use, but limited
experience with it. Sixty-two percent of respondents reported being very familiar or familiar
with IDPH’s Genetics and Newborn Screening Program.

Respondents had the opportunity to provide comments on many topics. They expressed
concern with insurance coverage and reimbursement issues, including constraints regarding
payment for genetic services; lack of physician education regarding genetics; the size and
distribution of the genetic provider workforce; and the need for public education. Many
concerns and uncertainties were described regarding the future of the field of genetics, notably
questions about the roles and responsibilities of trained genetic specialists and other non-genetic
health professionals. Respondents felt that IDPH should play a role in genetic services funding,
coverage and reimbursement issues, and education.

Interview Findings. Genetic service providers identified a number of key issues affecting
the current and future provision of genetic services in Illinois, including what they perceive to be
key needs and barriers with respect to care. These issues, which were identified repeatedly
throughout the interviews, fall readily into the categories of finance/reimbursement, workforce
adequacy, education, engagement, access, and a cluster of issues that have come to be known as ethical, legal, social issues or implications (ELSI). To summarize, from the perspective of genetic service providers, the issues that need to be addressed in the state include:

1. An inadequate and uneven system of third party reimbursement for genetic testing and clinical services.
2. The discrepancies between what are considered essential components of clinical care in genetics and what third party payers consider reimbursable services.
3. An inadequacy in the current supply of genetic professionals, including medical geneticists and genetic counselors.
4. A lack of effective integration between current systems of non-genetic primary and specialty care and genetics.
5. Knowledge, educational, and informational gaps and needs throughout the primary care and non-genetic health workforce.
6. Knowledge and informational gaps and needs among the public.
7. Access disparities, including (a) an uneven distribution of genetic service provision, and (b) a lack of linguistic and cultural diversity among genetic service providers.
8. Ethical, legal, and social concerns, including issues related to genetic discrimination and concerns that genetics may exacerbate existing or create new health disparities.

As key stakeholders in the genetic services delivery system, genetic service professionals provided insights into the current state of services in Illinois, anticipated future needs in the state, and the key barriers to care. The major themes that emerged were: the need to address access disparities, the need to address the inadequacies in finance/reimbursement, and the need to address knowledge deficits among the public and the primary care and larger health workforce. These also represent the most frequently noted barriers to care, and are highlighted by the role they suggested for the state department of public health, which focused on education, direct service provision, assuring access, and assuring a stable and secure system of finance/reimbursement for genetics. While emphasizing important systemic issues, genetic service providers also stressed the need for the state to continue to support public health’s role in the provision of direct services.

Local Health Departments

Survey Results. Of the 71 health departments that had a staff member respond to the survey, 50 provided some level of genetic services; 39 of those received grants from IDPH. Among the health departments that provided genetic services, there was variation between those that did and did not receive IDPH grant funding. A larger proportion of funded health departments provided genetic screening, on-site genetic clinics, referrals to genetic providers, and public and professional genetics education; a larger proportion also reported that they were able to meet their clients’ needs. None of the non-funded health departments had a genetic
coordinator on staff. When asked about the future impact of genetics on their public health programs, a greater proportion of funded than non-funded health departments foresaw a large impact, regardless of their provision of genetic services. Funded health departments were more comfortable with providing a variety of different services than were their non-funded counterparts, a finding that reveals areas where health department staff education can be targeted. The large proportion of respondents who did not know what public health programs areas would need to incorporate genetics in the next 3-5 years also indicates a need and opportunity for educational interventions. When asked about key issues that providers in local health departments will face over the next 5 to 10 years with respect to the provision of genetic services, respondents stressed the need for public education regarding genetic services. Other challenges included: incorporating genetics into chronic disease programs; program funding within the context of state budget constraints; inadequacies in insurance coverage; limited local access to genetic services; ethical, legal, and social issues, and non-genetic health care providers’ limited knowledge about genetics. They indicated that IDPH should play a role in funding genetic services (counseling), health department staff education, and community/public education.

Interview Findings. The challenges and access barriers identified by interviewees from local health departments appear to reflect the population served by local health departments (generally, poor and uninsured, often rural) as well as broader system issues: transportation problems; client non-compliance; lack of client education/awareness about genetics and genetic services; and financial/insurance constraints. Local health department interviewees also reported a need for more staff training in and awareness of genetics. The most frequently mentioned roles for IDPH were education (public, caregivers, health department staff) and funding (genetic testing, genetic services, transportation assistance).

Non-Genetic Health Care Providers

This small, diverse group of primary and specialty care providers provided insight into their current involvement with the provision of genetic services in Illinois, the challenges they face, and their thoughts on IDPH’s role in the genetic services system. Genetics has impacted all of their practices and is expected to continue to do so. However, non-genetic health care providers reported uncertainty about their place in the genetic service system. They were concerned about insurance and reimbursement issues, education for themselves and the public, and the supply of genetic service providers. Their recommendations for IDPH’s role in the genetic service system included service provision (i.e., genetic screening), public and health provider education, and financial support.

State Agency Personnel

Excluding those participants from the state’s genetic program, most interviewees from state agency programs reported having only minimal genetic knowledge and training and reported that their program areas included genetics only in limited ways. Interviewees discussed the need for program and agency staff to have greater awareness and education about genetics. They noted that, in addition to staff education, organizational structure and funding streams were barriers to integrating genetics into their programs. Overall, state agency staff indicated that IDPH’s role should be to coordinate and ensure access to genetic services, educate and provide
information to health care providers and the public about genetics, and to assume various data and surveillance functions.

**Consumer/Advocacy Organizations**

In this study sample, the focus of persons in the role of organizational leader or parent tended toward personal issues and the need for support services; how well health care providers communicate with families about genetic issues; and concerns about the accuracy of the information provided by health care professionals. Respondents in the role of advocacy professional tended to focus on education and training for themselves and for health professionals, as well as on the need for policies to protect the privacy of genetic information. Continuing education for clinicians and health care providers was already a service provided by two of the advocacy groups.

**Public Attitudes and Perceptions**

Surveying the public directly was beyond the scope of this needs assessment process. Our method for gauging attitudes and perspectives about ethical, legal, and social issues in genetics was to study secondary sources. Many studies are referenced in Chapter 7. The planning process will elicit input from Illinois residents on these vital issues.

**Illinois State Laws and Legislation**

Illinois has enacted several pieces of legislation related to genetics health policy over the last several years, including the Genetic Information Privacy Act of 1997 (GIPA) that broadly protects the confidentiality of genetic information, and the recent licensing of genetic counselors. The General Assembly has demonstrated willingness to consider important issues in the field.

**Discussion and Next Steps**

Four major themes were identified during the needs assessment process:

- limited genetic provider supply;
- low genetic literacy on the part of the general public;
- inadequate third party reimbursement; and
- lack of integration between genetics and the overall health care system.

The resulting public health issues will come before the IDPH as concerns requiring the department’s direct involvement, as well as partnership with other organizations, agencies and entities.

The overall challenge for IDPH is to identify, and assist others in identifying, the system in which all players are likely to be acting in the near and long-term future, and to articulate the performance standards and goals of such a system. In consultation with many stakeholders, the department can clarify which issues it wishes to directly address, and which it will influence through leadership. Illinois’ public health genetic services plan will provide a process for setting priorities among the many possible routes of action suggested by this needs assessment.
Chapter 1: Background

Genetics in the 21st century is about all diseases. Therefore, the new genetics will eventually change the face of public health by focusing interventions on individuals and groups who will benefit the most from behavioral modification, drug therapies, and other approaches to risk reduction. Genetics is now a fundamental tool of public health that will be integrated into public health programs.

--Muin J. Khoury MD, PhD, Director, Office of Genomics and Disease Prevention, CDC.1

In the next three or four years there’s going to be an absolute outpouring of discoveries about gene variances that are associated with the risk of diabetes, heart disease, cancer, asthma, high blood pressure, mental illness and other conditions. It will allow us to individualize programs of preventive medicine so that you could plan your own diet and lifestyle and medical surveillance based upon your genetic risks as opposed to some broad generic prescription of activities, which is what we currently do.

- Dr. Francis Collins, Director of the National Human Genome Research Institute.2

In the United States, public health genetics has been defined as “the application of advances in genetics and molecular biotechnology to improve public health and prevent disease.”3 This definition includes the broader public health goal of fulfilling society’s interest in assuring conditions in which people can be healthy.4 More current definitions of genetics and public health are starting to include the terminology “genomics and public health.” The term “genomics” refers to the study of the functions and interactions of all the genes in the genome, including their interactions with environmental factors.5

The origins of genetics and public health began with the study of the epidemiological basis of genetic disease. The early statistical models then gave rise to more clinical applications that involved population screening for specific genetic diseases, such as Tay Sachs and Sickle Cell. Later, in the 1960’s, prenatal diagnosis for chromosomal abnormalities and biochemical conditions became available.6 With the sequencing of the human genome, even more opportunities are available to identify, predict and prevent conditions with a genetic component.

To address both the opportunities and challenges posed by these advances on public health practice, in 1997 the Centers for Disease Control and Prevention (CDC) established an Office of Genetics and Disease Prevention and began developing a strategic plan. The CDC’s strategic plan encompassed four public health functions: 1) public health assessment; 2) evaluation of genetic testing; 3) development, implementation and evaluation of population interventions; and 4) communication and information dissemination.7 In addition to these four public health functions, three other critical issues were addressed within each function. The first

---

critical issue includes forging partnerships and coordinating efforts between various agencies and consumers. An example of this type of partnership has occurred between exchanges during genetics and public health conferences held by CDC, the Health Resources and Services Administration (HRSA), and the Association of State and Territorial Health Officials (ASTHO).8

The second critical issue identified by the CDC strategic plan is the recognition that the information gained from the human genome project will have profound ethical, legal and social implications for individuals and society.9 Some examples of these critical issues include informed consent in public health genetic research, population access to clinical and preventive services, privacy concerns in population-based surveillance programs and group stigmatization.10 In response to these concerns, the National Institutes of Health and Department of Energy Working Group on the Ethical, Legal and Social Implications of Human Genome Research has developed guidelines to promote safe and effective genetic testing in the United States.11

The third critical issue described in the CDC strategic plan is the recognition that more education and training of public health professionals will be needed to successfully integrate genetics into public health activities. In response to this issue, the CDC has published standard genomics competencies for the public health workforce.12 These competencies aim to help the public health practitioner apply genomic knowledge and tools within their professional responsibilities.

In summary, the field of genetics, or genomics and public health, will be growing immensely in the next decade. Contemporary public health genetics will involve integrating genetic knowledge and public health concepts to address a variety of conditions including many common chronic diseases (such as heart disease, diabetes and cancer), infectious diseases, and environmental health. Methods of integrating the new technology in ways that will benefit the health of individuals and society will continue to challenge this exciting new field.

Purpose

With the completion of the Human Genome Project in 2003, there has been an increase in scientific, media and public interest in genetics. The CDC articulated a vision of public health in the 21st century that emphasizes the integration of genetics/genomics into public health practice. As a part of this vision, the CDC has encouraged every state to develop a genetics state plan identifying the specific ways in which states will address their current and future genetic service and policy needs. The first step in the process of developing a state plan is the completion of a needs assessment.

The purpose of this needs assessment is to assess the role and function of the state health department with regard to current and future needs relating to genetic policy, service provision,

---

10 Centers for Disease Control and Prevention, 1997.
workforce and community education, and research and surveillance in Illinois. This was accomplished through:

- An assessment of the current provision of genetic services in Illinois; and,
- An assessment of the essential elements of a state genetics program, currently and in the coming years, defined through the perspectives of key stakeholders in Illinois, and the visions and practices of key state and national public health agencies.

The needs assessment will serve as the foundation for the development of a state public health genetic services plan.

**Brief overview of methods**

The needs assessment process involved the review of key documents and program literature, as well as surveys and interviews conducted with key stakeholders throughout the state. A complete description of the survey and interview methods is found in Appendix 1. Surveys were sent to genetic coordinators or maternal and child health coordinators at all 94 local health departments (see Appendix 2 for questionnaire) and to all identified genetic service providers in Illinois, including medical and laboratory geneticists, genetic counselors, and genetics nurses (see Appendix 3 for questionnaire). A total of 86 interviews were conducted with genetic service providers, other health care providers, local health department personnel, state agency staff, and representatives of advocacy groups (see Appendix 4 for interview guides).
Chapter 2: Illinois State Profile

Understanding current demographics, diversity, economic outlook, and health indicators in Illinois is important for planning genetic health services and programs. The following overview provides a context for considering current and future genetic resource and service needs.

Geographic and Demographic Overview

Geographic Density and Distribution

Covering approximately 56.5 thousand square miles, Illinois is the 24th largest state in the country by geographic area, and the fifth largest by population with an estimated 12,713,634 people.\(^\text{13}\) The population density for the state is 225 persons per square mile – almost three times the national average. As indicated in Figure 2.1, Illinois’ population is primarily concentrated in the northeast corner of the state, in and around the Chicago metropolitan area.

**Figure 2.1 Illinois Population Density, 2000\(^\text{14}\)**

---


The Chicago-Naperville-Joliet Metropolitan Statistical Area (hereafter Chicago MSA), which includes Cook and seven other surrounding or “collar” counties in the northeastern part of the state, accounts for 7.8 million residents, or more than 60 percent of the state’s total population. The Chicago MSA includes the first and second largest cities in the state: Chicago, with slightly over 2.8 million residents and Aurora with 166,614 residents. The third largest city in the state, Rockford, with a population of 152,452, is located approximately 90 miles to the west of Chicago, in Winnebago county. Outside of the Chicago-Naperville-Joliet and Rockford metropolitan areas, the state’s population density is highest around the metropolitan areas of Rock Island-Moline, on the northwest border with Iowa; Peoria, Bloomington-Normal, Springfield, Champaign-Urbana, and Charleston-Mattoon, in the central part of the state; St. Louis Metroeast area (East St. Louis, Belleville, Alton) in the southwest corner of the state; and Carbondale-Marion in the south.

Of Illinois’ 102 counties, 36 are considered metro (or urban) and 66 are considered non-metropolitan (or rural) according to the 2003 Office of Management and Budget (OMB) revised metro-nonmetro classification scheme. According to OMB definition, metro areas include central counties, with population thresholds of 50,000 or more residing in urbanized areas, and/or adjacent outlying counties, with high degrees of economic integration. Nonmetro areas, thus, represent counties that do not meet the 50,000 urbanized population threshold and/or the criteria for definition as an “outlying” county. The distribution of metropolitan counties approximates the population density as represented in Figure 1 above. According to 2004 estimates, approximately 1.7 million people (13.3 percent of the state population) live in counties that are considered rural (or nonmetro) and slightly over 11 million (86.7 percent of the state population) live in counties that are considered metro (or urban).

---


population) live in urban (or metro) counties in Illinois.\textsuperscript{19} Over two thirds of the state’s population resides in either the Chicago or St. Louis Metroeast metropolitan statistical areas.\textsuperscript{20}

\textbf{Population Diversity}

According to 2003 Census Bureau estimates, approximately 80 percent of Illinois’ population is white, 16 percent is Black or African American, slightly over 4 percent is Asian, six tenths of a percent is Native American, and one tenth of a percent is Native Hawaiian and/or Pacific Islander.\textsuperscript{21,22} Approximately 1 in 7 residents are of Hispanic/Latino origin.

Of the state’s Hispanic/Latino population, it is estimated that the majority, 78 percent, are of Mexican heritage. Slightly over 8 percent are estimated to be of Puerto Rican heritage, slightly over 4 percent to be of South American heritage, 3 percent to be of Central American heritage, one percent to be of Cuban heritage, and less than one percent to be of Dominican heritage.\textsuperscript{23}

\begin{itemize}
\item \textsuperscript{21} In this section, racial and ethnic categories represent those defined by the U.S. Office of Management and Budget (OMB) and utilized by the U.S. Census Bureau. The OMB requires federal agencies to utilize a minimum of two ethnicities (“Hispanic or Latino” and “Not Hispanic or Latino”) and five racial categories (“White,” “Black or African American,” “American Indian and Alaska Native,” “Asian,” and “Native Hawaiian and Other Pacific Islander”) in data collection and reporting activities. See, United States. Office of Management and Budget. Tabulation Working Group of the Interagency Committee for the Review of Standards for Data on Race and Ethnicity. “Provisional Guidance on the Implementation of the 1997 Standards for Federal Data on Race and Ethnicity,” December 15, 2000 <http://www.whitehouse.gov/omb/inforeg/re_guidance2000update.pdf>; United States. Office of Management and Budget. “Revisions to the Standards for the Classification of Federal Data on Race and Ethnicity.” October 30, 1997 (Volume 62, Number 210). <http://www.whitehouse.gov/omb/fedreg/1997standards.html> Though utilizing the minimum race and ethnicity categories as defined by the OMB, the 2000 decennial census included subcategories and was the first U.S. census that allowed respondents to select one or more racial categories to represent their racial identity. For a discussion of U.S. Census 2000 categories of race/ethnicity see <http://www.census.gov/prod/2001pubs/c2kbr01-1.pdf>\textsuperscript{22}
\item \textsuperscript{22} The American Psychological Association (APA) \textit{Publication Manual} recommends capitalizing names of colors when they are used to refer to human groups. See \url{http://www.apastyle.org/race.html}. Accessed January 26, 2006.
\end{itemize}
Table 2.1 2003 Illinois Population Proportions, by Race Alone or in Combination and Hispanic/Latino Heritage

<table>
<thead>
<tr>
<th></th>
<th>Non Hispanic/Latino</th>
<th>Hispanic/Latino</th>
<th>Total</th>
</tr>
</thead>
<tbody>
<tr>
<td>White</td>
<td>66.6</td>
<td>12.9</td>
<td>79.5</td>
</tr>
<tr>
<td>Black or African American</td>
<td>15.1</td>
<td>0.4</td>
<td>15.5</td>
</tr>
<tr>
<td>American Indian &amp; Alaskan Natives</td>
<td>0.4</td>
<td>0.2</td>
<td>0.6</td>
</tr>
<tr>
<td>Asian</td>
<td>4.2</td>
<td>0.1</td>
<td>4.3</td>
</tr>
<tr>
<td>Native Hawaiian &amp; Other Pacific Islanders</td>
<td>0.1</td>
<td>0.0</td>
<td>0.1</td>
</tr>
<tr>
<td>Total</td>
<td>86.4</td>
<td>13.6</td>
<td>100</td>
</tr>
</tbody>
</table>

Of the state’s Asian population, approximately 30 percent are estimated to be of Indian heritage, 21 percent of Filipino heritage, 17 percent of Chinese (except Taiwanese) heritage, 15 percent of Korean heritage, 4 percent of Japanese heritage, and 3 percent of Vietnamese heritage. According to 2004 estimates, 66.8 percent of the state’s Hispanic/Latino population and 56.4 percent of the state’s Asian population reside in Cook County.

The 2000 census indicated a foreign-born population of more than 1.5 million people in the state; 12 percent of the state’s total population. This represents a 63 percent increase from the 1990 census count. About half of individuals identifying as foreign-born are from Latin America and approximately one quarter are from Europe or Asia. Only very small numbers, relatively speaking, come from outside these three areas. About 40 percent of the total foreign-born population is from Mexico, 9 percent is from Poland, and almost 6 percent is from India. All other nationalities take up less than 5 percent of the total foreign-born population.

Population Growth

Illinois’ population grew almost 9 percent, or by about 1 million people, between 1990 and 2000; 2004 estimates show the state has grown over 2 percent in the first part of this decade. A large part of this growth results from an increase in the Hispanic/Latino population in Illinois from 904,446 in 1990 to 1,530,262 in 2000 (a 69 percent increase). The Asian population also saw significant growth, increasing 68 percent during the same time period. Individuals of Mexican heritage accounted for 84 percent of the growth in the Hispanic/Latino population during the 1990s, and individuals of Indian heritage accounted for 43 percent of the growth.

---

24 Adapted from U.S. Census Bureau (2004), Annual Estimates of the Population by Race Alone or in Combination; Vintage 2003. Available at [http://www.census.gov/popest/states/asrh/SC-EST2003-03.html](http://www.census.gov/popest/states/asrh/SC-EST2003-03.html). Accessed: June 1, 2005. This table incorporates individuals reporting one or more races. The 2000 Census was the first to allow respondents to identify with one or more racial categories. In 2000, 98.1 percent of the population of Illinois identified themselves with only one race, with 1.9 percent identifying with two or more races.


the Asian population during the same decade. Mirroring nationwide trends, between 2000 and 2003 the population of persons reporting Hispanic or Latino heritage of any race increased 12 percent, and the population of persons reporting Asian ancestry increased by nearly 14 percent in Illinois (Figure 2.2).

Figure 2.2 Illinois Population Growth by Race Alone or in Combination and Ethnic Origin, 2000-2003

The foreign-born population in Illinois increased by 63 percent between 1990 and 2000. Individuals from Mexico, Poland, and India accounted for much of the increase. The foreign-born Mexican population increased 121 percent during the 1990s, representing close to 335,000 people. The number of foreign-born residents from Poland increased 67 percent, by more than 56,000 people, and the number from India increased 125 percent, representing over 48,000 people. The number of state residents from the former Soviet Union, Pakistan, and Guatemala also more than doubled during the 1990s.

Almost 90 percent of the state’s growth between 1990 and 2000 was in the Chicago area, concentrated primarily in the suburban counties. In Figure 2.3, shaded counties represent the state’s ten fastest growing counties between 1990 and 2000; all but one – McLean County in

28 Adapted from U.S. Census Bureau, Annual Estimates of the Population by Race Alone or in Combination; Vintage 2003. Available at http://www.census.gov/popest/states/asrh/SC-EST2003-03.html
central Illinois – are in the northeastern corner of the state. The population in the suburban counties surrounding Chicago grew 25.5 percent between 1990 and 2000, and 11 percent between 2000 and 2004. In the city of Chicago and Cook County, the population grew by 4 and 5 percent respectively between 1990 and 2000, but declined by approximately 1 percent between 2000 and 2003.

**Figure 2.3 The Ten Fastest Growing Illinois Counties, 1990-2000**

Between 1990 and 2000, 34 counties in Illinois lost population; all but three of these counties were rural counties. However, tracking trends across this variable can be difficult because population growth in rural counties located near urban centers will shift the definition of the county into metropolitan status (and vice versa). Nine counties were re-categorized from their 1993 rural status as metropolitan for 2003; only one county was re-categorized as non-metropolitan. This led to an immediate 9% decrease in the total numbers of rural residents living in Illinois.29 Table 2.2 below shows that the proportion of rural residents has been steadily decreasing since 198030.

29Governor’s Rural Affairs Council. (2004). *Building on rural assets for a better future.* Available at [http://www.state.il.us/ltgov/pdf/GRAC_2004.pdf](http://www.state.il.us/ltgov/pdf/GRAC_2004.pdf). Accessed May 1, 2005. Metropolitan counties have close social and economic ties to counties containing large urban areas; all other counties are non-metropolitan, or rural. For more information on the definition of metropolitan and rural counties, see [http://www.census.gov/geo/www/ua/ua_2k.html](http://www.census.gov/geo/www/ua/ua_2k.html)

30Governor’s Rural Affairs Council. (2004). *Building on rural assets for a better future.* Available at [http://www.state.il.us/ltgov/pdf/GRAC_2004.pdf](http://www.state.il.us/ltgov/pdf/GRAC_2004.pdf). Accessed May 1, 2005. Metropolitan counties have close social and economic ties to counties containing large urban areas; all other counties are non-metropolitan, or rural. For more information on the definition of metropolitan and rural counties, see [http://www.census.gov/geo/www/ua/ua_2k.html](http://www.census.gov/geo/www/ua/ua_2k.html)
Table 2.2 Change in the Illinois Distribution of Population by Metropolitan/Rural, 1980-2004

<table>
<thead>
<tr>
<th>Year</th>
<th>% of Total, Rural</th>
<th>% Change, Rural</th>
<th>% of Total, Metropolitan</th>
<th>% Change, Metropolitan</th>
</tr>
</thead>
<tbody>
<tr>
<td>1980</td>
<td>15.5</td>
<td>--</td>
<td>84.5</td>
<td>--</td>
</tr>
<tr>
<td>1990</td>
<td>14.7</td>
<td>-5.3</td>
<td>85.3</td>
<td>1.0</td>
</tr>
<tr>
<td>2000</td>
<td>13.7</td>
<td>1.5</td>
<td>86.3</td>
<td>9.9</td>
</tr>
<tr>
<td>2004 (estimates)</td>
<td>13.3</td>
<td>-0.9</td>
<td>86.7</td>
<td>2.9</td>
</tr>
</tbody>
</table>

Economic Outlook

Like many states with large, urban areas, Illinois has a majority of its population and economic resources concentrated in a relatively small area. The median household income in Illinois is $46,590. About two-thirds of Illinois counties have median household incomes under $40,000, and eight counties, all from the southern tip of the state, have median household incomes below $30,000 (Figure 2.4). All rural Illinois counties have median incomes below the state median.

The unemployment rate in the state, at 5.9 percent for April 2005, is slightly above the national average of 5.2 percent. Illinois’ workers in the manufacturing industry were hit particularly hard during the recent economic downswing. The state had the highest number of mass layoffs in the nation during 2003, but overall unemployment has been declining since then. Although unemployment itself does not vary much according to urban/rural status, there are several trends in rural employment that present special challenges for long term planning. Employment growth in rural areas tends to be in low paying service sectors, which is increasing the wage gap between urban and rural workers. Urban workers now make more than $8,500 in wages per job more than rural workers. As high paying jobs become more concentrated in urban areas, the state may witness an increase in rural to urban migration. Additionally, several rural areas in the state experienced manufacturing plant closings. In rural areas, these closings have a broader and more long-term effect on the population, because displaced workers have fewer avenues to choose from in order to find new jobs.

Unemployment affects the Black or African American population in Illinois far greater than it affects the White population. The annual unemployment rate for 2002 was 6.5 percent; 6 percent of Whites were unemployed compared to 12 percent of Blacks and 8 percent of Hispanics/Latinos.\(^{36}\)

Current estimates show that 13 percent of the Illinois population lives in poverty; this represents a substantial increase from the 2000 level of 11 percent and is now above the most recent high of 12 percent recorded in 1990. Of the state’s impoverished residents, 39 percent are African American, 26 percent are Hispanic/Latino, and 11 percent are White.\(^{37}\) About 17 percent of children under 18 currently live in poverty, 46 percent of whom are African American, 22 percent are of Hispanic/Latino heritage, 11 percent are White, and 10 percent are Asian.\(^{38}\)

---


Twenty percent of children living in poverty have moved within the past 12 months, more than double the rate for non-impoverished children.\(^{39}\)

The U.S. Department of Agriculture estimates that in 2002, the poverty rate for rural counties was 12 percent and for metropolitan counties, it was 11 percent. However, Illinois residents in poverty still are disproportionately living in rural counties. Nearly 17 percent of the state’s poor live in rural areas, although just over 13 percent of the total population lives in rural areas. Although the state’s overall rural poverty rate declined during the 1990s, seventeen rural counties had their poverty rates increase by more than 6 percent.\(^{40}\)

Poverty affects the rural counties in the southern tip of the state more than it does the rural counties in the central, northwest, and west. Ten counties in the southern part of the state have child poverty rates over 20 percent. Three-quarters of the counties that had poverty rates above the state average of 11 percent in 2000 were in the southern and/or southwestern part of the state.\(^{41}\)

Figure 2.5 shows that although poverty affects highly urbanized areas like Chicago, the metropolitan suburbs and smaller metropolitan areas tend to have lower poverty rates than rural areas. And, as indicated by counties shaded white, while metropolitan suburbs can have relatively low poverty rates, they may still need to contend with large numbers of the poor because of their size.

---

\(^{39}\) National Center for Children in Poverty; available at [http://nccp.org/state_detail_demographic_IL.html](http://nccp.org/state_detail_demographic_IL.html)


The percentage of people living in Illinois without health insurance has risen dramatically over the past 15 years – by approximately 42 percent since 1988; the total portion of the population under the age of 65 without health insurance is now 16 percent. The proportion of individuals without health insurance is greatest in the Chicago area, where nearly one in four residents is uninsured. The suburban counties and the rest of the state both have averages under 15 percent.\(^{43}\)

The state’s percentage of uninsured children is slightly below the national average; nationwide, 11 percent of children were uninsured in 2003 compared to just 10 percent in Illinois. Nearly 1 in Illinois’ residents between the ages of 19 and 29 do not have health insurance, compared to 17 percent of persons aged 30-49 and 12 percent of persons aged 50-64.

Children without private health insurance and whose families meet income requirements may be enrolled in either Medicaid or KidCare (Illinois’ State Children’s Health Insurance Program, or SCHIP). Children in families earning up to 133 percent of the federal poverty level (FPL) are eligible for Medicaid and those in families earning between 133- 200 percent of the FPL are eligible for SCHIP. More than 92,000 children were insured through SCHIP in December 2003, a 20 percent increase from the December 2002 totals.\(^{44, 45}\) Just under 1 million children are enrolled in Illinois’ Medicaid plans. Figure 2.6 below shows the proportions of children under 18 insured by private and public insurance, as well as those without insurance. Children from families who earn too much to qualify for either Medicaid or KidCare will be able to enroll in a new comprehensive health insurance program, All Kids, which will begin July 1, 2006.\(^{46}\)

Figure 2.6 Illinois Insurance Status, Ages 0-18\(^{47}\)

Medicaid enrollment totals for 2003 were more than 1.7 million individuals; this represents an average monthly enrollment increase of 8.5 percent.\(^{48}\) Estimates for 2004 show that

---


\(^{44}\) Kaiser Family Foundation. Kaiser statehealthfacts.org

\(^{45}\) Illinois KidCare is the state’s SCHIP organization for insuring low-income children. Eligibility is set at 200% of the federal poverty level, currently $37,701 for a family of four.


\(^{47}\) Gilead Outreach and Referral Center. Numbers & Neighbors: A Detailed Description of Illinois’ Uninsured.
nearly 1 in 6 people in the state is enrolled in Medicaid.\textsuperscript{49} The state has no public insurance available for childless adults without disabilities, although parents of children enrolled in KidCare and pregnant women may also be insured through KidCare, if they meet the income requirements. More recently, the state has moved towards increasing enrollment of children through Medicaid, and Illinois currently is among state leaders in increasing the proportion of children insured through public programs.

Health insurance in rural areas of Illinois presents several challenges. Nationally, rural workers are less likely than urban workers to receive health insurance benefits.\textsuperscript{50} Medicaid enrollment in rural counties is higher than the rest of the state. Of the fifteen Illinois counties with Medicaid enrollments over 20 percent of the population, thirteen were in the southern half of the state.\textsuperscript{51} Rural counties tend to have lower proportions of individuals without health insurance of any type, perhaps because of greater Medicaid enrollment rates. Additionally, rural counties have a higher proportion of elderly residents – who may be insured through Medicare.\textsuperscript{52}

Managed care penetration in Illinois, at 13 percent, is well below the national average of 24 percent.\textsuperscript{53} Enrollment in managed care plans continues to decline – at a 7 percent rate of decline during 2003 and 3 percent rate during the first half of 2004. However, profits for the managed care plans in the state are rising, and financing experts feel that enrollment in these plans may well rebound.\textsuperscript{54,55} Only 11 percent of state Medicaid recipients were enrolled in managed care plans in October 2004, compared to nearly 60 percent rate nationwide.\textsuperscript{56}

Racial and ethnic disparities exist in the uninsured population, with Whites being uninsured at an 11 percent rate, Blacks at a 25 percent rate, and Hispanics/Latinos at a 30 percent rate. About 18 percent of Hispanics/Latinos who are U.S. citizens are uninsured compared to 57 percent of Hispanics/Latinos who do not have U.S. citizenship.

The uninsured are more likely to be from lower income families and tend to have less education. Forty-eight percent of families making under $25,000 have at least one family member who is uninsured, compared to 29 percent of families making $25-50,000, 22 percent of families making $50-75,000, and 15 percent of families making over $75,000. Of working persons, 16.0 percent are uninsured, including 15.6 percent of those working full-time. Nearly 80 percent of all uninsured persons in Illinois have at least one family member who is working.\textsuperscript{57} As education levels increase the uninsured rate decreases with 42 percent of people without high school diplomas uninsured compared to 19 percent of people with a high school diploma but without a college degree, and 9 percent of people with college degrees.

\textsuperscript{48} Kaiser Family Foundation. Kaiser statehealthfacts.org
\textsuperscript{49} Rich, R. F. “Medicaid, Managed Care, and SCHIP: Challenges and Issues” Presented at New Legislator’s Conference 2004; Springfield, IL. Available at: www.igpa.uiuc.edu/events/newMember/rich.ppt
\textsuperscript{53} Kaiser Family Foundation. Kaiser statehealthfacts.org. Includes all HMO and POS clients, public and private
\textsuperscript{54} Highlights of Illinois Managed Care Review 2004. Available at http://www.allanbaumgarten.com/index.cfm?fuseaction=dsp_report&state=il
\textsuperscript{55} Although managed care penetration in decreasing in the private sector, it is actually increasing in the public sector: between 2003-2004, Medicaid managed care enrollment increased 18.6%. Rich, R. F. Medicaid Managed Care and SCHIP.
\textsuperscript{56} Ibid.
Vital Statistics

There were 182,393 births in Illinois during 2003. Although annual variation has been very slight, long-term trends indicate that the birth rate is declining. The number of live births has shown an annual increase in the state just three times since 1990. Women between the ages of 20-34, who account for about 75 percent of all births, currently make up about 10 percent of the total population (Table 2.3). This population decreased about 8 percent between 1990 and 2000 and has remained moderately stable since, contributing to overall lower levels of fertility in the state since 1990. The state fertility rate, calculated as the number of live births per 1,000 women aged 15-44, has declined from 72.6 births per 1000 in 1990 to 67.4 births per 1000 in 2000.

Table 2.3 2004 Illinois Population Proportions, by Age and Sex

<table>
<thead>
<tr>
<th></th>
<th>&lt;10</th>
<th>10-19</th>
<th>20-34</th>
<th>35-64</th>
<th>&gt;65</th>
<th>Total</th>
</tr>
</thead>
<tbody>
<tr>
<td>Male</td>
<td>7.1</td>
<td>7.3</td>
<td>10.9</td>
<td>18.9</td>
<td>4.9</td>
<td>49.1</td>
</tr>
<tr>
<td>Female</td>
<td>6.8</td>
<td>7.0</td>
<td>10.4</td>
<td>19.6</td>
<td>7.1</td>
<td>50.9</td>
</tr>
<tr>
<td>Total</td>
<td>13.9</td>
<td>14.3</td>
<td>21.3</td>
<td>38.5</td>
<td>12.0</td>
<td>100</td>
</tr>
</tbody>
</table>

While births to White and Black women decreased substantially during the 1990s, births to Asian and Hispanic/Latino women increased substantially (Table 2.4). Births to women over the age of 35 years, the age associated with an increased risk of chromosome aneuploidy, increased 45 percent, from 17,929 in 1990 to 25,911 in 2000. Births to women over 35 years of age varies, however, with race/ethnicity, with White women representing 17 percent of births to women age 35 or older, and Hispanic/Latino and Black women representing 9 percent of births respectively.

Table 2.4 Percent of Live Births by Race & Ethnicity, 1990-2002

<table>
<thead>
<tr>
<th>Year</th>
<th>Total Births</th>
<th>White, Not Hispanic/Latino</th>
<th>Black, Not Hispanic/Latino</th>
<th>American Indian</th>
<th>Asian/Pacific Islander</th>
<th>Hispanic/Latino</th>
</tr>
</thead>
<tbody>
<tr>
<td>2002</td>
<td>180,622</td>
<td>55.0%</td>
<td>17.5%</td>
<td>0.1%</td>
<td>4.6%</td>
<td>22.7%</td>
</tr>
<tr>
<td>2001</td>
<td>184,064</td>
<td>55.2%</td>
<td>17.9%</td>
<td>0.1%</td>
<td>4.4%</td>
<td>22.3%</td>
</tr>
<tr>
<td>2000</td>
<td>185,036</td>
<td>55.8%</td>
<td>18.4%</td>
<td>0.1%</td>
<td>4.4%</td>
<td>21.2%</td>
</tr>
<tr>
<td>1995</td>
<td>185,812</td>
<td>59.3%</td>
<td>20.1%</td>
<td>0.1%</td>
<td>3.1%</td>
<td>17.3%</td>
</tr>
<tr>
<td>1990</td>
<td>195,790</td>
<td>62.7%</td>
<td>21.4%</td>
<td>0.1%</td>
<td>2.5%</td>
<td>12.4%</td>
</tr>
</tbody>
</table>

60 Ibid.
61 Adapted from U.S. Census Bureau, Annual Estimates of the Population by Sex and Age for Illinois. Available at http://www.census.gov/popest/statessr/SC-est2004-02.html
The state’s 2002 infant mortality rate was 7.2 per 1000. Infant mortality varies widely by race; the rate for Black infants has been two to three times larger than the rate for White infants since at least 1980 (Figure 2.7). In 2002, rates were 5.5 for whites, 5.8 for Hispanics/Latinos, and 15.7 for Blacks. Infant mortality statistics for Hispanics/Latinos are closer to whites than Blacks. The perinatal death rate for 2000 was 11.8 deaths per 1000; again, the rate of perinatal deaths for Blacks, at 20.7 per 1000, was more than twice the rate for whites (9.9 per 1,000).

Figure 2.7 Illinois Infant Mortality Rates by Race/Ethnicity, 1992-2002

Figure 2.8 below shows the county-by-county perinatal death rate in the state during the years 1998-2002. The overall perinatal death rate for this five-year period was 118.3 per 10,000. Of note is general correlation between Figures 2.8 and 2.1, indicating that more densely populated counties in the state have higher rates of perinatal death.

---


63 IDPH Center for Health Statistics. Vital Statistics Illinois 2000. Perinatal death rate is defined as deaths occurring in fetuses between 20 weeks estimated gestation and 7 days postnatal age. This contrasts with infant mortality, defined as deaths of live-born babies within the first year of life.

64 Illinois Kids Count 2005.
Newborn Screening

The newborn screening for genetic diseases and newborn hearing screening programs, and related data, are described in the current status section.

Birth Defects and Adverse Pregnancy Outcomes

Birth defects in Illinois are tracked via the Adverse Pregnancy Outcomes Reporting System, or APORS. The system tracks the incidence and geographic location of a number of birth defects and adverse pregnancies outcomes in the state. Table 2.5 below shows the total number of cases tracked by APORS and annual averages of those cases for the period, 1998-2002 (see also the discussion of the APORS system in Chapter 3). It should be noted that while APORS represents the most comprehensive source for birth defect and adverse pregnancy outcome information in Illinois, it is a passive surveillance system, reliant primarily upon hospital report, representing cases identified and reported by hospitals during or within a week following the newborn hospitalization period. Cases identified and/or diagnosed at a later time (e.g., when an infant is 6 or 14 months of age) are not included. It has been recognized that such a limited time frame for reporting birth defects and other adverse pregnancy outcomes most likely contributes to systematic underreporting of these outcomes, as many conditions are not readily identifiable during the early newborn period. Therefore, in the following discussion, it should be remembered that the incidence rates generated from the APORS data may underestimate the true rates of adverse pregnancy outcomes in Illinois.

---

66 Unless, that is, the infant has remained hospitalized through six or fourteen months of age, in which case the outcome would be reported.
Nearly 45% of birth defects (106.7 per 10,000 live births) tracked by the APORS system are cardiovascular defects. While serious and potentially fatal, many of these defects can be repaired surgically shortly after birth.

Central nervous system defects occur less frequently but have fewer procedural treatments and can cause severe mental retardation or death. According to APORS, the rate of central nervous system defects in the state is 12.4 per 10,000 live births; however, this number may underestimate the actual incidence because it does not track pregnancies not carried to term due to spontaneous pregnancy loss or elective termination of a fetus prenatally diagnosed with, for example, an open neural tube defect.

Table 2.5 Frequency of Infants Meeting APORS Case Criteria, 1998 – 2002

<table>
<thead>
<tr>
<th>Infants</th>
<th>5-Year Total</th>
<th>Annual Average</th>
<th>Rate (per 10,000 live births)</th>
<th>% APORS Cases</th>
</tr>
</thead>
<tbody>
<tr>
<td>Total APORS Cases</td>
<td>85,147</td>
<td>17,029.4</td>
<td>931.5</td>
<td>100.0</td>
</tr>
<tr>
<td>Intensive Care &gt; 24 hours (meets no other APORS criteria)</td>
<td>37,883</td>
<td>7,576.6</td>
<td>414.4</td>
<td>44.5</td>
</tr>
<tr>
<td>Birth Defects</td>
<td>20,653</td>
<td>4,130.6</td>
<td>225.9</td>
<td>24.3</td>
</tr>
<tr>
<td>Very Low Birth Weight</td>
<td>17,574</td>
<td>3,514.8</td>
<td>192.3</td>
<td>20.6</td>
</tr>
<tr>
<td>Positive for Controlled Substances</td>
<td>7,252</td>
<td>1,450.4</td>
<td>79.3</td>
<td>8.5</td>
</tr>
<tr>
<td>Fetal Deaths</td>
<td>6,267</td>
<td>1,253.4</td>
<td>68.6</td>
<td>7.4</td>
</tr>
<tr>
<td>Congenital Infections</td>
<td>4,853</td>
<td>970.6</td>
<td>53.1</td>
<td>5.7</td>
</tr>
<tr>
<td>Died During Newborn Hospitalization</td>
<td>4,547</td>
<td>909.4</td>
<td>49.7</td>
<td>5.3</td>
</tr>
<tr>
<td>Intrauterine Growth Retardation</td>
<td>2,557</td>
<td>511.4</td>
<td>28.0</td>
<td>3.0</td>
</tr>
<tr>
<td>Retinopathy of Prematurity</td>
<td>2,246</td>
<td>449.2</td>
<td>24.6</td>
<td>2.6</td>
</tr>
<tr>
<td>Endocrine, Metabolic, or Immune Disorder</td>
<td>264</td>
<td>52.8</td>
<td>2.9</td>
<td>0.3</td>
</tr>
<tr>
<td>Blood Disorder</td>
<td>193</td>
<td>38.6</td>
<td>2.1</td>
<td>0.2</td>
</tr>
<tr>
<td>Fetal Alcohol Syndrome</td>
<td>170</td>
<td>34.0</td>
<td>1.9</td>
<td>0.2</td>
</tr>
<tr>
<td>Other Conditions69</td>
<td>162</td>
<td>32.4</td>
<td>1.8</td>
<td>0.2</td>
</tr>
</tbody>
</table>

Chromosomal anomalies arise from abnormal numbers of chromosomes or from breaks, deletions or duplications in specific parts of chromosomes. Chromosomal anomalies have variable effects, causing death, mental retardation, malformations, or deformations depending on the condition and its severity. The most common chromosomal anomaly at birth is Down syndrome, which appears at a rate of 10.6 per 10,000 live births. The overall rate of chromosomal anomalies identified through APORS in the state is 13.6 per 10,000 live births.

68 Illinois Department of Public Health Division of Epidemiological Studies. (2004). Birth defects and other adverse pregnancy outcomes in Illinois, 1998-2002. A Report on County-Specific Incidence. Available at: http://www.idph.state.il.us/about/epi/pdf/report02.pdf. Accessed May 31, 2004. The numbers in this table do not denote an actual count of infants. In tabulating birth defects, the same baby may be counted multiple times because the baby may have multiple birth defects within even the same birth defect group. For example, a baby born with two multiple endocrine, metabolic, or immune disorders would be counted twice on the appropriate line.

69 Neurofibromatosis, chorioretinitis, strabismus, endocardial fibroelastosis, occlusion of cerebral arteries, cerebral lipidoses.
Alimentary tract defects as classified within the APORS system include orofacial and gastrointestinal defects such as cleft lip, cleft palate and esophageal atresia. Many have surgical corrections; they are identified through APORS in about 17.9 live births per 10,000 in the state.

Genitourinary defects affect the male and female external genitalia and reproductive systems and the renal system. The most common defects can often be surgically corrected, however some abnormalities, such as absence or severe malformation of the kidneys, frequently cause death within the first few hours of life. The state rate for all genitourinary defects is 22.0 per 10,000 live births.

Musculoskeletal defects may include problems like clubfoot, congenital hip dislocation, limb reduction defects, and omphalocele — a protrusion of the intestine through the abdominal wall and into the umbilical cord. The severity of these disorders is variable; a variety of corrections may or may not be available for each particular defect. The rate of these disorders as identified through APORS is 19.1 per 10,000 live births in the state.

Birth defects involving the respiratory system are also less common than those affecting other organs. The most frequently occurring is lung agenesis/hypoplasia, an underdevelopment of one or both lungs. It is identified by APORS at a rate of 2.8 per 10,000 live births in the state.

APORS data for congenital endocrine, metabolic, and immune disorders are inconsistent with data from the newborn screening program, since infants with these conditions are often diagnosed after the newborn hospitalization period. Therefore, data for these conditions are instead reported in the following chapter in the discussion of the Newborn Screening Program.

**Children with Special Health Care Needs**

DSCC is the Illinois Title V Program for Children with Special Health Care Needs. DSCC provides care coordination, benefits management and financial assistance for diagnostic and treatment services to children (through age 21) who meet financial eligibility and have, or are suspected of having, a treatable chronic medical condition in one of the following categories:

- Orthopedic conditions (bone, muscle, joint disease)
- Heart defects
- Hearing loss
- Neurological conditions (nerve, brain, spinal cord)
- Certain birth defects
- Disfiguring defects such as cleft lip, cleft palate, and severe burn scars
- Speech conditions which need medical/dental treatment
- Certain chronic disorders such as hemophilia and cystic fibrosis
- Certain inborn metabolic problems including phenylketonuria (PKU) and galactosemia
- Eye impairments including cataracts, glaucoma, strabismus and certain retinal conditions - excluding isolated refractive errors
- Urinary system impairments (kidney, ureter, bladder)

Through cooperative agreement with IDPH, DSCC covers the cost of diagnostic testing as needed for infants with specifically identified positive or suspect newborn screens. Children with conditions and/or impairments that meet DSCC’s medical eligibility criteria may also receive continued services through DSCC based on financial eligibility. It is important to note

---

that DSCC’s eligibility criteria are primarily impairment, not diagnosis, based. This means that although, for example, children with endocrine disorders or Down Syndrome (as opposed to cystic fibrosis, for example) may not be eligible for services on the basis of their diagnosis, they are eligible for services related to impairments falling within the categories of medical eligibility. Currently, impairments associated with gastrointestinal, hematology, endocrine, and pulmonology conditions are not medically eligible for service through DSCC.\(^71\) DSCC serves approximately 20,000 families from all of Illinois’ 102 counties annually through 13 regional offices\(^72\).

In 2002, the National Survey of Children with Special Health Care Needs (CSHCN) estimated the total number of CSHCN in Illinois at 385,269 children, or 11.7 percent of children under 18 years of age.\(^73\)

Tables 2.6-2.8 below summarize the insurance status and access to health services of these children in Illinois.

### Table 2.6 Health Insurance Coverage for Illinois CSHCN

<table>
<thead>
<tr>
<th>Health Insurance Coverage</th>
<th>State %</th>
<th>National %</th>
</tr>
</thead>
<tbody>
<tr>
<td>Percent of Children with Special Health Care Needs without insurance at some point in the past year.</td>
<td>10.5</td>
<td>11.6</td>
</tr>
<tr>
<td>Percent of Children with Special Health Care Needs currently uninsured.</td>
<td>4.2</td>
<td>5.2</td>
</tr>
<tr>
<td>Percent of currently insured Children with Special Health Care Needs with insurance that is not adequate.</td>
<td>41.4</td>
<td>33.5</td>
</tr>
</tbody>
</table>

### Table 2.7 Access to Healthcare for Illinois CSHCN

<table>
<thead>
<tr>
<th>Access to Care</th>
<th>State %</th>
<th>National %</th>
</tr>
</thead>
<tbody>
<tr>
<td>Percent of Children with Special Health Care Needs with any unmet need for specific health care services.</td>
<td>16.9</td>
<td>17.7</td>
</tr>
<tr>
<td>Percent of Children with Special Health Care Needs with any unmet need for family support services.</td>
<td>4.3</td>
<td>5.1</td>
</tr>
<tr>
<td>Percent of Children with Special Health Care Needs needing specialty care who had difficulty getting a referral.</td>
<td>26.7</td>
<td>21.9</td>
</tr>
<tr>
<td>Percent of Children with Special Health Care Needs without a usual source of care (or who rely on the emergency room).</td>
<td>10.6</td>
<td>9.3</td>
</tr>
<tr>
<td>Percent of Children with Special Health Care Needs without a personal doctor or nurse.</td>
<td>11.0</td>
<td>11.0</td>
</tr>
</tbody>
</table>

\(^{71}\) Information provided in interview.

\(^{72}\) To find an office, see http://internet.dssc.uic.edu/dsccroot/office_lookup.asp

\(^{73}\) HRSA Maternal Child Health Bureau. (2001). *National Survey of Children with Special Health Care Needs Chartbook. Illinois State Data*. Available at [http://mchb.hrsa.gov/chscn/state_data/il.htm](http://mchb.hrsa.gov/chscn/state_data/il.htm). Accessed: May 24, 2005. The survey was conducted by the CDC’s National Center for Health Statistics. Data are self-reported by parents. Although the survey tracked more racial and ethnic variables than reflected here, the survey suppresses small results to protect the privacy of the sample. Thus, some ethnic/racial data is not available.
Table 2.8 Socioeconomic Impact on Families with CSHCN

<table>
<thead>
<tr>
<th>Impact On Family</th>
<th>State %</th>
<th>National %</th>
</tr>
</thead>
<tbody>
<tr>
<td>Percent of Children with Special Health Care Needs whose families pay $1,000 or more in medical expenses per year.</td>
<td>10.4</td>
<td>11.0</td>
</tr>
<tr>
<td>Percent of Children with Special Health Care Needs whose condition caused financial problems for the family.</td>
<td>22.0</td>
<td>20.9</td>
</tr>
<tr>
<td>Percent of Children with Special Health Care Needs whose families spend 11 or more hours per week providing or coordinating care.</td>
<td>13.4</td>
<td>13.5</td>
</tr>
<tr>
<td>Percent of Children with Special Health Care Needs whose condition affected the employment of family members.</td>
<td>28.8</td>
<td>29.9</td>
</tr>
</tbody>
</table>

The state is comparable to national averages for many of these variables. Exceptions include the percentage of parents who feel their child’s health insurance is not adequate and the percentage of CSHCN who had difficulty getting a referral; the state is noticeably above national averages for these variables.

Nationally, CSHCN are more likely to have public insurance and less likely to be uninsured than other children. CSHCN from low-income families are more likely to be uninsured than those from higher income families. The data show that despite public insurance initiatives, families still must spend large amounts of time, money, and energy to ensure their children’s basic health care needs are met. Nearly 1 in 3 of CSHCN affected the employment of family members, but more than 1 in 6 still have an unmet need for a specific health care service.

Adult Genetic Conditions and Chronic Diseases with Genetic Components

Traditionally, public health genetics has focused on newborn screening and treatments for pediatric patients. This trend is beginning to change as the genetic factors in many common chronic conditions that cause high levels of morbidity and mortality in adults are increasingly recognized and understood. With an increasing understanding of the genetic etiologies of complex conditions, such as cancers, heart disease, Alzheimer’s disease, diabetes, bipolar disorders, and schizophrenia, among others, and the increasing availability of genetic information about and testing options for such conditions, the demand on public health to integrate genetics into its chronic disease initiatives and ensure adequate information and service provision is expected to grow. It is expected that the genetic components of these and many other chronic diseases will continue to be discovered in the years to come, leading to a continually increasing demand for adult genetic services statewide.

---

Cancer

Nationally, cancer causes more than 20 percent of all deaths and is one of the only leading causes of death that has not declined during the last 50 years.\textsuperscript{75} In Illinois, cancer causes approximately 25,000 deaths per year and represents the second leading cause of death in the state, behind heart disease.\textsuperscript{76} It is the leading cause of death for all individuals between the ages of 35 and 74.\textsuperscript{77} Genes and heredity are thought to contribute to individual susceptibility in multiple cancers. The discussion here focuses on breast and ovarian cancer and colorectal cancers, due to their incidence rates, availability of tests, and identified genes.

Breast/Ovarian Cancer.\textsuperscript{78} Breast cancer is the most commonly diagnosed cancer in Illinois women, accounting for nearly 1 in 3 cancer diagnoses. It causes about 2,000 deaths per year in Illinois, and represents the second leading cause of cancer deaths among women. Though less prevalent in men, male breast cancer in the state accounts for another 70 cases and 20 deaths annually. The age-adjusted breast cancer incidence rate\textsuperscript{79} in Illinois has increased sharply since the mid 1980s – from an incidence rate of 65.2 in 1986 to 72.8 in 2001, for both sexes, and from 115.5 to 131.8 in 1986 and 2001 respectively, for women.\textsuperscript{80} The rate of diagnosis for breast cancer \textit{in situ} is also increasing, from 10.0 in 1986 to 30.7 in 2001.\textsuperscript{81} At the same time, mortality rates for women have decreased, from 35.6 in 1986 to 26.5 in 2001. For men, the mortality rate from breast cancer has increased from 0.3 in 1986 to 0.5 in 2001. It is estimated that in 2005, more than 9,000 persons in Illinois will be diagnosed with and over 1900 deaths will result from breast cancer. Approximately 99 percent of these projected cases are expected to relate to female breast cancer.

The incidence rates of breast cancer are higher for White woman than for woman of all other races and ethnicities. Mortality rates, by contrast, are highest for Black women. Between 1986 and 2001, the average incidence rate in Illinois was 114.9 for Black women and 130.2 for White women, while the average mortality rate was 40.0 for Black women and 32.1 for White women. While breast cancer mortality rates decreased in Illinois between 1986 and 2001 for White and Black women, the rate of decrease for White women was much greater (from 35.0 to 25.4 for White women, compared with 42.3 to 37.2 for Black women). The incidence and mortality rates for breast cancer are lower for Hispanic/Latino women than for non-Hispanic/Latino women, with Hispanic/Latino women in Illinois having an incidence rate of 85.8 in 2001 versus an incidence rate of 134.9 for non-Hispanic/Latino women, and a mortality rate of 12.4 versus 27.0 for non-Hispanic/Latino women. Mortality rates have declined since 1990 for both Hispanic/Latino and non-Hispanic/Latino women, however the decrease has been greater for non-Hispanic/Latino women.

\textsuperscript{76} Illinois Department of Public Health. Vital Statistics. Available at: \url{http://www.idph.state.il.us/health/statshome.htm}.
\textsuperscript{79} Except where otherwise indicated, all rates given in this section are per 100,000 and are age adjusted to the U.S. standard million population.
\textsuperscript{80} For men, the age-adjusted breast cancer incidence rate increased from 1.1 in 1986 to 1.3 in 2001.
\textsuperscript{81} \textit{In situ} breast cancers are localized, non-invasive, pre-malignant tumors, i.e. cancers that have not yet spread from their originary ducts or lobules, and as such tend to have more favorable prognoses.
Ovarian cancer, accounting for approximately 4 percent of all women’s cancers, represents the fourth leading cause of cancer mortality for women. The high mortality rate associated with ovarian cancer (the highest for all cancers of the female reproductive system) is thought to result in part from a lack of early symptoms and effective screening systems.\(^{82}\) Between 1998 and 2002, ovarian cancer caused on average 611 deaths per year in Illinois (an annual death rate of approximately 8.9).\(^{83}\) Between 1986 and 2001, the incidence rate of ovarian cancer in Illinois averaged 14.8 per 100,000, declining somewhat from 14.7 in 1986 to 12.8 in 2001. The incidence rate is higher for Whites, than for women of all other races, and for non-Hispanic/Latinos compared with women of Hispanic/Latino heritage. Between 1986 and 2001, mortality rates from ovarian cancer decreased for White women and women not of Hispanic/Latino heritage. For Black women, mortality rates increased between 1998 and 2001, with Black women having the highest mortality rate (9.1) from ovarian cancer in 2001.

It is estimated that 5 to 10 percent of all breast and ovarian cancers are hereditary.\(^{84}\) American women with a first degree relative with breast cancer are 2.1 times more likely than other women to develop breast cancer themselves, and relative risk is inversely correlated with the age of the relative’s diagnosis. Similarly, women with a single first degree relative with ovarian cancer are more than three times as likely to develop ovarian cancer themselves. For women with 2 or 3 first-degree relatives, the relative risk increases to nearly 5 times the likelihood of developing ovarian cancer. There is some evidence to suggest that women with a first degree relative with ovarian cancer are at a modest increased risk for breast cancer as well.\(^{85}\)

Of the genetic mutations known to be associated with increased risks of breast and ovarian cancer, the most common are the BRCA1 and BRCA2 genes. While current estimates of lifetime risk suggest that 13.2 percent of all women in the population will develop breast cancer, for women who carry the known BRCA1 and BRCA2 mutations, the lifetime risk estimates range from 36 to 85 percent. Women carrying either of these genes have up to an 85 percent chance of developing breast cancer before their 80\(^{\text{th}}\) birthday. Similarly, while lifetime risk estimates suggest that ovarian cancer will affect 1.7 percent of women in the general population, women with the BRCA1 and/or BRCA2 mutations face risk estimates as high as 60 percent.\(^{86}\) There is hope that continued discoveries concerning the role of genetics in breast and ovarian cancers will help reduce morbidity and mortality from these diseases.

Colorectal Cancers. About 7,000 cases of colorectal cancer are diagnosed in Illinois annually. It is the cause of about 2,500 deaths in the state annually, and represents the third leading cause of cancer deaths among both men and women. Colon cancers are both more common and more deadly than rectal cancers: the incidence rates are 44.1 for colon cancer and 16.1 for rectal cancers, while the mortality rates are 22.1 for colon cancer and 3.3 for rectal cancers. In Illinois,\(^{87}\) the incidence and mortality rates for colorectal cancers are higher for men

---


\(^{83}\) National Cancer Institute, State Cancer Profiles, Illinois, http://statecancerprofiles.cancer.gov/


than women. For men, the incidence rates for colon and rectal cancers are 51.4 and 21.4 respectively; for women, the incidence rates are 39.2 per 100,000 and 12.3 per 100,000 respectively. Mortality rates are 1.5 to 2 times higher for men than for women. Consistent with national trends, Blacks in Illinois have the highest incidence and mortality rates related to colon cancer. In 2001, the age-adjusted incidence rate for colon cancer among Blacks in Illinois was 51.8 compared with 40.7 among Whites and 30.8 among Asians and other races. The age adjusted mortality rates from colon cancer in 2001 were 27.5 among Blacks, 18.7 among Whites, and 11.8 for Asians and other races. Non-Hispanics/Latinos are 2.5 to 3 times more likely to be affected by colorectal cancers than Hispanics/Latinos.

The absolute risk for an American to develop colon cancer before age 79 is about 1 in 25. A family history positive for colorectal cancer can greatly increase these odds. One first degree relative raises the risk approximately 2.5 times; more than one first degree relative with colorectal cancer, or a single first degree relative diagnosed before age 45 increases the risk approximately 4 times. It is estimated that approximately 5 to 10 percent of colorectal cancers have a genetic etiology. Genes contributing to susceptibility to two types of colon cancer, familial adenomatous polyposis (FAP) and hereditary nonpolyposis colorectal cancer (HNPCC), have been identified. HNPCC, the more common of the diagnoses, accounts for approximately 2 percent of all cases of colorectal cancer. To date, mutations in four genes have been linked to HNPCC. Individual carriers of these mutations are estimated to have a lifetime risk of 80 percent for developing colon cancer. FAP is an inherited syndrome in which hundreds of polyps form in the colon and rectum. FAP, which accounts for less than one percent of all colorectal cancers, has been linked thus far with over 300 different mutations in one gene, the APC gene. It is estimated that individuals who have inherited mutations in this gene have a near 100 percent chance of developing colorectal cancer by the time they are 40 years of age.

Heart Disease

Heart disease is the leading cause of death nationally and in Illinois. Heart disease accounts for approximately 30,000 deaths in Illinois annually, nearly 30 percent of total deaths in the state. Mortality rates for both men and women from heart disease are higher in Illinois, 251.1 per 100,000, than nationally, 246.8 per 100,000. Blacks are much more likely than other racial groups to die from heart disease, with a 334.6 per 100,000 mortality rate; the

---


corresponding rate for whites is 242.1 and for all other races, it is just 126.7. The figure for Blacks is well above the national average of 315.8 per 100,000. Rates for the state’s Asian and Hispanic/Latino populations are well below national averages.

Heart disease deaths are more likely to occur downstate than in the Chicago area. Mortality rates in the southern third of Illinois are above the state average of 536 per 100,000 (Figure 2.9). Most of the central portion of the state has rates under the state average, and the more populous northern third is a mix.

**Figure 2.9 Illinois Total Population 35+, Heart Disease Death Rates 1996-2000**

According to Behavioral Risk Factor Surveillance System (BRFSS) survey results in 2003, adults in Illinois reported the following risk factors for heart disease and stroke:

- 24 percent had high blood pressure
- 34 percent of those screened reported having high blood cholesterol
- 7 percent had diabetes
- 23 percent were current smokers

---


94 National Center for Chronic Disease Prevention and Health Promotion, Division of Adult and Community Health. (2005). Interactive Cardiovascular Health Maps. Available at [http://apps.nccd.cdc.gov/giscvh/(pprnsvu53g3jpi551p10isur)/default.aspx](http://apps.nccd.cdc.gov/giscvh/(pprnsvu53g3jpi551p10isur)/default.aspx). Accessed: June 16, 2005. Exact rates for Asians and Hispanics/Latinos compare slightly different data - only the population over age 35. Additionally, the CDC’s data set is older, with the most recent year measured being 2000 instead of 2003.

95 CDC Interactive Cardiovascular Health Maps. Because rates are shown only for population age 35 and over, they are higher than the statehealthfacts.org figures, which include total population.

• 61 percent were overweight or obese (Body Mass Index greater than or equal to 25.0)
• 26 percent reported no exercise in the prior 30 days
• Approximately 83 percent had one or more of these six risk factors.

Several genetic causes of heart disease have been identified and are currently being researched. For example, dilated cardiomyopathy is a disorder that has been linked to several gene loci. Several genetic studies have identified “susceptibility loci” for coronary artery disease which, along with the complication of acute myocardial infarction, is a leading cause of morbidity and mortality in developed countries.”

Alzheimer’s Disease

Alzheimer’s disease is a condition with a complex etiology. More than 211,000 Illinois residents currently have Alzheimer’s disease. It now accounts for more than 2,000 deaths in the state annually – a more than 200 percent increase since the early 1990s. It is estimated that 1 in 10 Illinois residents aged 65 or older and almost half of the population over 85 years of age is affected by Alzheimer’s disease. Although the nation is expected to see an average 44 percent increase in Alzheimer’s cases by the year 2025, the projected increase in Illinois is much lower, about 14 percent, primarily because the population growth in the state’s elderly population is projected to be slower than the national growth. However, this 14 percent increase still represents nearly 30,000 cases of Alzheimer’s disease in Illinois during the next 20 years.

Current estimates suggest that between 5 and 25 percent of cases are hereditary. There are two recognized forms of Alzheimer’s disease: early onset familial Alzheimer’s Disease and late onset Alzheimer’s disease. Early onset Alzheimer’s disease, defined in part by the appearance of symptoms prior to age 65, tends to run in families, and represents less than 5 percent of all incidents of Alzheimer’s disease. Mutations in three genes have been found to be causally related to early onset Alzheimer’s, including the $\beta$-amyloid (A$\beta$) precursor protein (APP) on chromosome 21, and the presenilin 1 (PSEN1) and presenilin 2 (PSEN2) genes on chromosomes 14 and 1 respectively. Late onset Alzheimer’s disease, which manifests after age 65, represents the majority of cases of Alzheimer’s disease. Late onset Alzheimer’s disease has a less determinate genetic etiology. Though genetics is thought to play a role in this form of Alzheimer’s, no specific genes have yet been identified as causal agents. One gene, however, the apolipoprotein E (APOE) gene, has been associated with an increased risk for developing late

onset Alzheimer’s disease. A number of other genes are suspected of playing a role in increased susceptibility to late onset Alzheimer’s disease, however research has yet to confirm any of these potential genetic associations.

**Diabetes**

Reflecting national trends, the incidence of diabetes is rapidly expanding in the Illinois. The IDPH estimates that more than 500,000 adults in the state have diabetes; according to national estimates, more than 1/3 of these individuals are undiagnosed and not under a physician’s care. Type 2 diabetes accounts for 90 – 95 percent of all cases nationwide; although typically adult-onset, there is an increasing incidence of children and adolescents with type 2 diabetes.

Although about 7 percent of the state is affected, the elderly and non-white ethnic groups are most likely to be affected. Almost 40 percent of those diagnosed with the condition in the state are over age 65 and more than 85 percent of the state’s cases are over age 45. African Americans are 1.6 times more likely to get diabetes than non-Hispanic/Latinos Whites, and Hispanics/Latinos are 1.5 times more likely to get diabetes than non-Hispanics/Latinos. Just over 8 percent of all Hispanics/Latinos in the U.S. have diabetes, while nearly 1 in 4 Mexicans between the ages of 45-64 have diabetes.

Diabetes is responsible for more than 3,000 deaths in Illinois annually – a 37 percent increase since the early 1990s. The death rate is 25.4 per 100,000 population. Men are more likely to die from diabetes than women (28.1 to 23.3 rate), and Blacks are much more likely to die than Whites and other racial groups (42.8 to 23.3 to 20.5, respectively).

**Mental Health**

Mental illness accounts for 4 of the top 10 cases of disability in the U.S. and affects over 44 million adults – more than 1 out of 5 Americans - in any given year. The Kaiser Family Foundation reports that 32 percent of Illinois residents have reported poor mental health during


110 National Institutes of Mental Health
the past 30 days – with women, Whites, and Blacks being more likely to report poor mental health than men and other racial/ethnic groups.\textsuperscript{111}

Schizophrenia and bipolar disorder, two of the more debilitating mental illnesses, both affect about 1 percent of Americans – indicating that 127,000 Illinois residents have these conditions.\textsuperscript{112} Both conditions have been shown to have complex gene/environment interactions; also, first degree relatives of people with bipolar disorder are 10-20 times more likely to develop a bipolar or depressive disorder themselves, and first degree relatives of people with schizophrenia are 6-10 times more likely to develop schizophrenia.\textsuperscript{113}

Summary

This chapter provides a detailed overview of Illinois population and health characteristics, including: geographic density and distribution; population diversity and growth; economic outlook; health insurance status; vital statistics; birth defects and adverse pregnancy outcomes; children with special health care needs; and adult genetic conditions and chronic diseases with genetic components (such as cancer and heart disease). These data are relevant to consider when planning for the future of the genetic services system in Illinois.


Chapter 3: Current Status of Public Health Genetics in Illinois

The Illinois Department of Public Health’s (IDPH) Genetics and Newborn Screening Program is the key coordinator of public health genetics in the state. The program has the following responsibilities: administering the state’s legally mandated universal newborn screening program; coordinating and overseeing a statewide public/private network of clinical genetic centers and local health departments; and providing educational programs to the public and health care providers on genetics and health promotion. In addition to IDPH, the Illinois Department of Human Services (IDHS), the administrator of the Title V Maternal and Child Health Block Grant, and the Division of Specialized Care for Children (DSCC), the state’s Title V Children with Special Health Care Needs Agency, provide case management, clinical care, and support services to women and children with reproductive and special health care needs, including those associated with certain types of genetic conditions and/or indications associated with genetic components or risk factors.

Illinois Department of Public Health, Genetics and Newborn Screening Program

The Illinois Department of Public Health’s Genetics and Newborn Screening Program is located within the Office of Health Promotion, Division of Health Assessment and Screening, along with the Sudden Infant Death Syndrome (SIDS)/Infant Mortality, Vision and Hearing, Childhood Lead Poisoning Prevention, and Perinatal Programs. The Genetics and Newborn Screening Program currently administers two core programs: the Newborn Screening Program and the Genetics Program. These programs along with other key activities are described below.

Newborn Screening Program

The Newborn Screening Program (NBS Program) was established in 1965 with the passage of legislation mandating universal newborn screening for phenylketonuria (PKU); which represented the first direct programmatic involvement in clinical genetic services by the state of Illinois. The program expanded over the next three decades to include five other inherited/metabolic disorders: congenital hypothyroidism (1979), galactosemia (1984), biotinidase deficiency (1986), congenital adrenal hyperplasia (1987), and sickle cell disease (1989). With the adoption of tandem mass spectrometry testing (MS/MS) in 2002, a technology that enables screening for a wide range of disorders from a single blood sample, IDPH expanded its newborn screening panel to include amino acid, fatty acid oxidation, and organic acid disorders. Illinois was one of the first states to implement universal expanded newborn screening through tandem mass spectrometry and in 2002 was one of only four states screening universally for over twenty disorders.\textsuperscript{114} Illinois currently screens for over 35 inherited metabolic disorders\textsuperscript{115} (see Appendix 5), and is considering the addition of cystic fibrosis to its newborn screening panel. It is estimated

\textsuperscript{114} United States General Accounting Office. (March 2003.) \textit{Newborn Screening: Characteristics of State Programs.} GAO-03-449, Appendix II, p 31. According to the GAO report, in 2002, while most states required universal screening for eight or fewer disorders, Illinois was screening for twenty-seven disorders. The other three states requiring universal screening for over twenty disorders (exact number of disorders is given in parentheses) at that time were: North Carolina (32), Oregon (33), and Wisconsin (21).

\textsuperscript{115} IDPH. (Feb 2005). Division of Laboratories. Manual of Services, p. 28-29, for a listing of disorders currently screened via newborn screening program. The Manual is available online at: http://www.idph.state.il.us/about/laboratories/labman.pdf
that universal newborn screening for cystic fibrosis will detect approximately 50 cases per year, given current birthrates, population rates of cystic fibrosis, and the racial/ethnic characteristic of the population in Illinois. The Genetic and Metabolic Diseases Advisory Committee, the oversight committee to IDPH Genetics and Newborn Screening Program, has submitted a recommendation to the IDPH to add cystic fibrosis to the state’s screening panel.\textsuperscript{116}

The NBS Program involves the collaborative efforts of hospitals and birthing facilities, the IDPH Newborn Metabolic Screening Laboratory, NBS Program staff, local health departments, private physicians, and parents. In accordance with legislative mandate, all hospitals, physicians, and midwives in the state of Illinois are responsible for acquiring blood samples by heel stick from all infants delivered under their care as soon as the infant is between 24 and 48 hours of age. Screening samples are submitted to the state’s Newborn Metabolic Screening Laboratory in Chicago, ideally within 24 hours of collection. The Newborn Metabolic Screening Laboratory is a state laboratory organizationally located within IDPH’s Division of Laboratories. It is the sole authorized facility for the analysis of mandated newborn screening samples. The Laboratory maintains a database of all samples screened and reports information on all screens to the submitting facilities. All abnormal or suspect screens are additionally reported to the NBS Program staff, who contact the physician of record on the specimen card to report the results of any abnormal screening. Typically the physician contacts the family and arranges for follow-up diagnostic and treatment services; however, in cases where the physician is unable to locate the family, local health department nurses may be asked to make contact with the family.

Follow up services, including further diagnostic testing, clinical care, and treatment services (e.g., medical products for newborns diagnosed with certain metabolic disorders and prophylactic medications for clients with sickling conditions), are also coordinated through, and provided by the NBS Program, in conjunction with local health departments and other public health agencies (e.g., DSCC). The Newborn Screening Program also maintains a database of information on all clients with positive newborn screens, tracking follow-up services, diagnostic, and developmental information for all newborns/children with a confirmed diagnosis through age 15 years (see below for more information on this and other relevant databases).

Over 180,000 infants are screened annually in Illinois through the newborn screening program. In 2004, the state newborn screening laboratory analyzed 183,727 specimens (including initial and repeat specimens), identifying 10,603 specimens requiring some degree of follow up service, with 259 genetic, metabolic, and/or endocrine disorders diagnostically confirmed.\textsuperscript{117} Over 5,200 confirmed diagnoses for metabolic or sickling disorders in infants have been identified through the newborn screening program since its inception. Table 3.1 presents the number of confirmed cases, by disorder/condition, identified through newborn screening as of 2004 (note that the year in parenthesis corresponds to the year screening was initiated):

\textsuperscript{116} Genetic and Metabolic Diseases Advisory Committee. Minutes—November 18, 2004 Meeting.
\textsuperscript{117} IDPH, Division of Health Assessment and Screening. Newborn Screening Follow-Up Services, 2003 Process Data and 2004 Preliminary Process Data. Note that as of March 25, 2005, several additional cases remained pending diagnosis or resolution.
Table 3.1 Cumulative Number of Confirmed Cases, March 2005

<table>
<thead>
<tr>
<th>Disorders/Conditions</th>
<th>Cumulative Number of Confirmed Cases</th>
</tr>
</thead>
<tbody>
<tr>
<td>Phenylketonuria (1965)</td>
<td>790</td>
</tr>
<tr>
<td>Congenital Primary Hypothyroidism (1979)</td>
<td>1,575</td>
</tr>
<tr>
<td>Galactosemia (1984)</td>
<td>350</td>
</tr>
<tr>
<td>Biotinidase Deficiency (1986)</td>
<td>21</td>
</tr>
<tr>
<td>Congenital Adrenal Hyperplasia (1987)</td>
<td>200</td>
</tr>
<tr>
<td>Hemoglobinopathies (1989)</td>
<td>2,2</td>
</tr>
<tr>
<td>Amino Acid Disorders (2002)</td>
<td>10</td>
</tr>
<tr>
<td>Fatty Acid Oxidation Disorders (2002)</td>
<td>63</td>
</tr>
<tr>
<td>Organic Acid Disorders (2002)</td>
<td>31</td>
</tr>
</tbody>
</table>

To ensure the timeliness of newborn screening sample delivery, IDPH currently supports a courier service that provides overnight sample delivery from hospitals and other facilities to the state’s Newborn Metabolic Screening Laboratory. Over 50 hospitals are currently participating in this program.119

The IDPH Newborn Screening Program supports follow-up services for infants identified as positive for a genetic/metabolic condition through the state’s screening program. For infants identified as positive for sickle cell disease or trait and/or other hemoglobinopathies, diagnostic testing, family counseling, and long-term medical management and care are supported through a pediatric hematology grant program, which provides funding to pediatric hematology centers located across the state.120 Initiated in 1989, in conjunction with the institution of screening for sickle cell disease, the program currently supports the services of eleven pediatric hematology centers, located in Chicago, Peoria, Urbana, St. Louis, and the East St. Louis (see Figure 3.1). In 2004, 786 patients received services through the grant funded pediatric hematologists121 and over 17,500 patients have received services since the program was initiated in 1989 through fiscal year 2004.122

---

118 Counts are from IDPH NBS Program, representing confirmed cases as of March 2005. Note that the counts are not exact and the number of total confirmed cases is potentially higher for each disorder. Note also that PKU is an amino acid disorder and since 2002 has been screened utilizing tandem mass spectrometry. Between July 2002 and April 2005, over 52 cases of PKU were identified through tandem mass spectrometry and these are included in the PKU (n=790) counts in the table, not with the counts for amino acid disorders.

119 Genetic and Metabolic Diseases Advisory Committee. Minutes—November 18, 2004 Meeting.


121 Number served provided by Karen Burget, IDPH Genetics and Newborn Screening Program, in email communication, July, 12, 2005.

Figure 3.1 Pediatric Hematology Genetic Grants and Outreach Site – FY 05

**Chicago Area**
Children’s Memorial Hospital  
John H. Stroger, Jr. Hospital of Cook County  
Mt. Sinai Hospital  
Rush University Medical Center  
University of Chicago  
University of Illinois, Chicago

**Peoria Area**
St. Francis Medical Center  
+Macon County Health Department

**Champaign Area**
Carle Clinic

**Southern Illinois Area & St. Louis, MO**
Southern Illinois Health Care – Centreville (Mother Child Center)  
St. Louis University – Cardinal Glennon Hospital  
Washington University – St. Louis Children’s Hospital  
+Genetic satellite clinic site

**Clinical Genetics Program**

Since 1983, IDPH’s Genetics and NBS Program has also supported a statewide network of clinical genetic centers through the administration of a clinical genetics grant program with the intent of assuring community level access to genetic services throughout the state. With funding from the U.S. Department of Health and Human Services, the Clinical Genetics Program began in 1983 with grant awards to five genetic centers based at major medical facilities in Chicago, Rockford, Springfield, Peoria, and Champaign. In 1989 the program was expanded to include funding to ten local health departments and nine pediatric hematology centers. Today a statewide network of clinical genetic care currently includes sixteen university-based clinical genetic centers, thirty-nine local health departments, and eleven pediatric hematology centers; in 2004, these grantees served 7,889, 5,634, and 786 clients respectively. Currently, the sixteen clinical genetic centers

---

124 IDPH. Genetics and Newborn Screening Program. Clinical Genetic Program Report.
are located in: Chicago metropolitan area (10), Rockford (1), Springfield (1), Urbana (1), Peoria (1), and St. Louis (2) (see Figure 3.2). Funded genetic centers represent almost all of the primary genetic centers serving Illinois residents (see Overview of Clinical Genetics).

**Figure 3.2 Clinical Genetic Grantees – FY05 And Outreach Sites**

**Chicago Area**

- Advocate Christ Medical Center
  - (Hope Children’s Hospital)
  - +Will County Health Department
- Advocate Medical Group – Lutheran Hospital
- Children’s Memorial Hospital
- John H. Stroger Hospital of Cook County
  - (Hektoen Institute)
- Loyola University Medial Center
- Illinois Teratogen Information Service
  - (ITIS) Reprogenetics Research, Inc.
- Rush University Center
  - +Copley Hospital – Aurora
  - +St. Joseph’s Hospital – Elgin
  - +Riverside Medical Center – Kankakee
- University of Chicago
  - +LaRabida Children’s Hospital
  - +Friends and Family Center
- University of Illinois at Chicago – Metabolic
  - +Rockford Memorial Hospital
  - +Springfield Department of Public Health
  - +Peoria University of Illinois Medical Center
  - +Lisle
  - +Advocate Christ Hospital
- University of Illinois at Chicago – OB/GYN
  - +Chicago Department of Public Health

**Rockford Area**

- Rockford Memorial Hospital

**Peoria Area**

- University of Illinois at Peoria
  - +LaSalle County Health Department

**Champaign Area**

- University of IL at Champaign

**Springfield Area**

- Southern Illinois University Medical Center
  - +Macoupin County Health Department
  - +Montgomery County Health Department

**Southern Illinois Area and St. Louis, MO.**

- St Louis University at Cardinal Glennon Hospital

---


127 Number served provided by Karen Burget for the IDPH Genetics and Newborn Screening Program in email communication, July, 12, 2005.
Clinical Genetic Centers

Grants to clinical genetic centers primarily support the provision of genetic counseling services to individuals referred through funded local health departments.\(^{128}\) Funded centers provide a full range of prenatal/reproductive, pediatric, and adult/cancer genetics services, although individual centers may differ in their foci.\(^{129}\) In addition to the sixteen primary sites, funded centers currently provide outreach services at twenty-five satellite clinics across the state, eleven of which are located at local health departments.\(^{130}\) Since 1984, more than 82,747 clients have received services through the funded genetic centers, increasing steadily from 600 clients in 1983 to 7,889 in 2004 (see Figure 3.3).

In 2004\(^{131}\), the 7,889 patients served had 9,159 patient visits and resided in 99 of Illinois’ 102 counties. Despite the geographical reach of the program, approximately 78% of clients served were residents of urban areas. Patients served included 4,941 prenatal and 2,948 non-prenatal patients. Approximately 44% of prenatal patients represented women between thirty-five and forty-one years of age, with advanced maternal age being the primary reason for referral for prenatal patients (accounting for 44% of all referred prenatal patients). Other common reasons for referral of prenatal patients included: family history of inherited conditions, abnormal ultrasounds, abnormal maternal serum alpha-fetoprotein (MSAFP), and teratogen exposure. For non-prenatal patients, the majority were nine years of age or younger (64%) with close to 50% being four years of age or younger. Common reasons for pediatric referrals included developmental delays and birth defects.

\(^{128}\) In some cases, the grants support the services of other genetics related personnel. For example, in fiscal year 2004 funding supported 3 dieticians in addition to 23 genetic counselors. See, IDPH. Division of Health Assessment and Screening, Genetics Program. *Illinois Genetic Counseling Grants Summary Report*. FY 2004. July 2003-June 2004.


\(^{130}\) IDPH, Genetics and Newborn Screening Program. *Clinical Genetic Program Report*. Distributed at Genetic and Metabolic Diseases Advisory Committee Panel Meeting, May 20, 2004. Number of local health departments hosting outreach clinics provided by email from K. Burget, IDPH Genetics and Newborn Screening Program, July 12, 2005. These figures represent services for fiscal year 2005. The number of outreach clinics changes somewhat from year to year with shifts in funding.

Approximately 81% of all patients seen in 2004 were female, reflecting the fact that 62% of all patients received prenatal services. Racially and ethnically, patient demographics resembled state demographics as reflected in the 2000 U.S. Census, although, based on reporting, clients served in the genetics centers were slightly less likely to be white (68% of patients at genetics centers versus 73% of the population statewide), slightly more likely to be Black (19% of genetics patients versus 15% of the population statewide), and were slightly more likely to be Hispanic or Latino (17% of genetics patients versus 12% statewide) (see Figure 3.4 and Figure 3.5).132

In addition to providing services, IDPH-funded clinical genetics centers work closely with IDPH-funded local health departments and provide educational presentations to health care providers and consumers (see below).

Local Health Departments

Through grants to local health departments, IDPH’s Genetics and Newborn Screening program seeks to increase local access to clinical genetic information and services through a coordinated community based system of screening, case finding, referral and education that operates as a part of the statewide clinical genetics network. In 2004, thirty-nine local health departments (see Figure 3.6) were funded to provide: (a) follow up, referral, tracking, and educational services to infants with a positive newborn screen test result, (b) screening, referral and education for a disorder with a genetic component utilizing a standardized genetic screening tool, and (c) consumer and professional education. Thus, at the local health department, genetic services include the provision of follow-up services as well as case finding and referral, the latter involving family history screening, evaluation, and, when indicated, referral to funded clinical genetic centers for further services. Eleven of the 39 IDPH-funded local health departments refer clients to on-site genetics clinics

132 IDPH. Division of Health Assessment and Screening, Genetics Program. Illinois Genetic Counseling Grants Summary Report, July 2003-June 2004. Note, however, that 10.24 percent of patients did not report information on race and 19.39 percent did not report information on Hispanic ethnicity.
staffed by genetic counselors and MD geneticists from the funded clinical genetic centers. Currently genetic clinics are offered at health departments in Jackson, Jefferson, Kankakee, Knox, LaSalle, Macon, Macoupin, Madison, Montgomery, Sangamon, and Will counties. These clinics operate once per month, once every other month, once every 3 months, or twice per year, depending upon the county (see Appendix 6: List of Funded health departments). Some of the 39 funded health departments also serve as lead agencies and subcontract to fund smaller health departments to provide genetic screening, referral, and follow up services. Through this two-tiered system, it is estimated that the genetic services are being offered in over 60 local health departments across the state.133

133 Genetic and Metabolic Diseases Advisory Committee, Minutes—November 18, 2004 Meeting.
Figure 3.6 Local Health Department Genetic Grantees and Covered Counties – FY05

Local Health Departments (covered counties)
*Genetics clinic sites
Bond County Health Department
Bureau County Health Department (Putnam)
Champaign County Public Health Department
   (Clark, Coles, Cumberland, Douglas, Ford, Iroquois, Vermilion, City of Champaign & Urbana)
Chicago Department of Public Health (City of Chicago)
Cook County Health Department (Suburban Cook County)
Crawford County Health Department
DuPage County Health Department
East Side Health District (Portions of St. Clair Co.)
Edgar County Health Department
Egyptian County Health Department (Gallatin, Saline, White)
Evanston City Health Department
*Fayette County Health Department (Clay, Effingham, Jasper, Lawrence)
Henderson County Health Department (Warren)
Henry County Health Department (Stark)
*Jackson County Health Department (Perry, Franklin-Williamson)
*Jefferson County Health Department
Jersey County Health Department
   (Calhoun, Greene, Jersey)
Kane County Health Department
Kankakee County Health Department
Knox County Health Department
*LaSalle County Health Department
   (Livingston)
*Macon County Health Department (DeWitt, Moultrie, Piatt, Shelby)
*Macoupin County Health Department
*Madison County Health Department
Marion County Health Department
McDonough County Health Department
McLean County Health Department
Mercer County Health Department
Monroe-Randolph Bi-County Health Department
*Montgomery County Health Department
Rock Island County Health Department
 Sangamon County Health Department (Brown, Cass, Logan, Mason, Menard, Morgan, Pike, Schuyler, Scott)
Southern Seven Health Department (Alexander, Hardin, Johnson, Massac, Pope, Pulaski,

*Springfield Department of Public Health
St. Clair County Health Department
Tazewell County Health Department

*Will County Health Department
   (Grundy, Kendall)
Winnebago County Health Dept. (Boone, Carroll, DeKalb, JoDaviess, Lee, Olge, Stephenson, Whiteside)

LHD Grantees (38) in yellow (46 counties) Covered Areas by Grantees (44 counties in blue) Areas not covered in white (15)

* Genetic Screening Tool used by 65 counties
The IDPH Expanded Genetic Screening Tool (see Appendix 7) is a key method of identifying clients at risk for genetic conditions and who could benefit from genetic services. Originally developed in 1998 by the Genetics and Newborn Screening Program, the screening tool was modified and expanded in 2004 to incorporate questions related to various chronic diseases including cancers, heart disease, diabetes, asthma, and arthritis (see discussion of IDPH Genomics and Chronic Disease Prevention Program below).134 IDPH-funded local health departments utilize the screening tool to evaluate each client’s risk for genetic/hereditary diseases or disorders, and the tool, in conjunction with associated risk algorithms, serves as the basis for determining a client’s need for possible referral to a clinical genetic center. The screening tool has primarily been incorporated into maternal child health programs such as WIC/Family Case Management, Family Planning, the Adverse Pregnancy Outcomes Reporting System (APORS), and the Prenatal Clinic programs; these programs are explained later in this chapter.135 Nurses and/or case managers ideally complete the screening tool with each client utilizing services through these programs. Funding to local health departments supports the position of a Genetic Coordinator136 (usually a public health nurse with training in genetics) who is responsible in part for reviewing the genetic screening forms and determining the need for follow up services and/or referral. In conjunction with the health department staff, the genetic counselor at the clinical genetic center, and the client, the genetic coordinator ensures appropriate referrals are made. Clients may be referred to a clinical genetic center, to the local health department’s on-site clinic, or to their primary care practitioner, depending upon the responses to the screening tool.

In 2004, 33,666 clients were screened utilizing the genetic screening tool (a 7 percent increase from 2003) and 5,634 received additional genetic services (a 1.8 percent decrease from 2003). Of these, 4,398 had positive indicators for a genetic center referral and 1,708 accepted a referral. Of those accepting the referral, 14 percent are known to have followed through with an appointment at the genetic center. There is, however, no system in place to track follow through at all the genetic centers, making it impossible to know with certainty the actual degree of patient follow through, or the reasons for not scheduling an appointment.137

Of all clients receiving genetic services at funded local health departments in 2004, approximately 61 percent were female, 78 percent were 9 years of age or younger, with 72 percent younger than 24 months, and 54 percent were referred through the Adverse Pregnancy Outcomes Reporting System (APORS) program (see discussion of APORS Program below). Only 15.5 percent of clients receiving genetic services were between 20 and 49 years of age. The most common reasons for referral included: single malformations, functional disorders, reproductive risk, exposure to teratogen induced anomalies, chromosomal syndromes, multiple congenital anomalies, and sickle cell trait.138

---

134 Screening tool process information from “Genetic Services in the Local Health Department” November 2002. IDPH, Genetics and Newborn Screening Program. Unpublished manuscript.
136 Currently, this is most often supported as a part time position. In some cases, the position is filled by a dedicated part time person, and in other cases the role of genetic coordinator is combined with other roles assumed by a full time public health nurse.
Racial and ethnic demographics of clients served resemble the demographic characteristics of the state population as a whole as reflected in the 2000 U.S. Census. Compared to statewide racial and ethnic characteristics, however, clients served in funded local health departments were slightly more likely to identify their race as Black (25 percent of clients served versus 15.1 percent of the population statewide) and to be of Hispanic/Latino ethnicity (14.3 percent of clients served versus 12.3 percent of the population statewide) (see Figures 3.7 and 3.8). By contrast, clients served were less likely to report their race as white (64 percent of clients served versus 73.5 percent of the population statewide), and to be non-Hispanic/Latino (70.1 percent of clients receiving services, versus 87.7 percent of the population statewide).

Since 1991, over 101,951 clients have been screened utilizing the genetic screening tool, with genetic services (including follow up and referrals) provided to more than 51,410. The number of clients served has steadily increased over the years, as evidenced in Figure 3.9.

---

A key aspect of the clinical genetics program is education. As a part of their funding, IDPH grantees provide medical genetic educational presentations and programming to consumers and professionals. In 2004, funded local health departments provided more than 459 educational presentations to 19,670 consumers and 2,340 health care professionals and clinical genetic centers provided 32 presentations to 979 consumers, and 310 presentations to 11,734 health care professionals. Pediatric hematology grantees also engage in educational activities, providing presentations to 1,877 health care professionals and 3,194 consumers in 2004. Since the inception of the genetics grant program, over 321,000 consumers and professionals have participated in grant sponsored educational programs.

In addition, the Genetics and Newborn Screening Program has, since 1987, conducted an annual weeklong training program in clinical genetics for public health nurses; currently, this program is offered in conjunction with March of Dimes Illinois Chapter’s Annual Perinatal Conference. Over 1,000 public health nurses have received training in clinical genetics through this program. The training program incorporates presentations by expert speakers, break-out sessions, case studies, and panel discussions and is designed to provide participants with basic and up to date information related to clinical genetics. Topics covered include modes of inheritance, genetics assessments for pediatric, prenatal, and adult clients, teratogens and birth defects, genetics and chronic disease, genetic screening, resources and services available, and legal and ethical issues with respect to clinical genetics. The training program is consistently

---


highly evaluated by participants and has been nationally recognized as a model program for educating the public health workforce.\textsuperscript{143}

Genomics and Chronic Disease Prevention Project

In the fall of 2003, the IDPH Genetics and Newborn Screening Program initiated the Genomics and Chronic Disease Prevention Project with the aim of enhancing the state’s public health infrastructure by incorporating genomic information into IDPH’s Chronic Disease Prevention Programs and vice versa. A Genomics Team was formed which includes the program leadership from IDPH’s genetics and chronic disease programs, including IDPH’s asthma, cancer control, cardiovascular health, arthritis, WISEWOMAN, and Alzheimer’s prevention programs. The Project is coordinated by a contracted genetic counselor working through the Genetics and Newborn Screening Program. Meeting on a semi-annual basis, the team has updated IDPH’s chronic disease health fact sheets to include genetic/genomic information where appropriate and revised the Genetics Screening Tool utilized by IDPH-funded local health departments to incorporate screening questions and risk algorithms specific to chronic disease. Following lengthy deliberation, questions and risk algorithms specific to hereditary cancers, cardiovascular disease, asthma, diabetes, and arthritis were added to the screening tool (see Appendix 7: Expanded Genetic Screening Tool). In the spring of 2005, the Expanded Genetic Screening Tool was delivered to all funded local health departments, which are currently in the process of incorporating it into their genetic screening programs.

Other Activities

Currently the Genetics and Newborn Screening program is participating in a Department of Health and Human Services, Health Resources and Services Administration funded Region 4 Genetics Collaborative Project. Through this project, Illinois’ Genetics and Newborn Screening program is working with the genetics programs in Indiana, Kentucky, Michigan, Minnesota, Ohio, and Wisconsin to improve regional genetics communication and collaboration with a view toward benefiting each state’s genetics program. Specific objectives of the project include: (1) implementing and improving state administered expanded newborn screening systems; (2) reducing access inequities, specifically those related to geographic distributions of services through exchange clinics, telemedicine, long distance consultation, and other practices, and (3) developing a regional plan for improving the public health genetics infrastructure.\textsuperscript{144}

Genetic and Metabolic Diseases Advisory Committee

The Genetics and Newborn Screening Program works in collaboration with an IDPH Director appointed advisory committee, the Genetic and Metabolic Diseases Advisory Committee (GMDAC). The GMDAC is comprised of 20 voting members, representing medical genetics, pediatrics, hematology, endocrinology, genetic counseling, local health department genetic coordinator, social work, laboratory services, and parent advocates. Ex-officio members include representatives from IDPH’s Genetics and Newborn Screening Program, Newborn


\textsuperscript{144} For a description of the Region 4 Genetics Collaborative see: http://genes-r-us.uthscsa.edu/resources/genetics/State%20Pages/region4.htm
Metabolic Screening Laboratory, Office of Women’s Health, and Chronic Disease Prevention Programs, as well as from the Division of Specialized Care for Children, advocacy organizations, and the Genetic Task Force of Illinois. Also represented among ex officio members are the fields of medical ethics, pediatrics, and medical genetics (see Appendix 8). The GMDAC serves as the advisory committee to both the Genetics and Newborn Screening Program and the IDPH Newborn Metabolic Screening Laboratory. Currently, the GMDAC has three standing committees: (1) Newborn Screening Laboratory, (2) Education, Family and Community Services, and (3) New Directions/Planning/Legislation.

Genetics and Newborn Screening Funding/Revenue

The activities of the Genetics and Newborn Screening Program are primarily funded by a Newborn Screening Fee. The fee, currently $47 per newborn screening sample, is billed directly to hospitals and other entities for each specimen submitted to the state laboratory. All genetic grant programs, including grants to local health departments, clinical genetic centers, and pediatric hematologists, are funded from the Metabolic Screening and Treatment Fund. For most states, newborn screening fees represent the primary funding source for newborn screening programs. According to a national survey of newborn screening programs conducted by the Government Accountability Office (GAO), newborn screening fees funded 64 percent of newborn screening program expenditures nationwide in 2001, with 13 states relying entirely on NBS fees to fund their programs.

Newborn Hearing Screening Program

Legislatively mandated universal newborn hearing screening became effective in Illinois on December 31, 2002. IDPH’s Vision and Hearing Program, located within the Division of Health Assessment and Screening, administers the Illinois Newborn Hearing Program in conjunction with the Illinois Department of Human Services and Division of Specialized Care for Children (DSCC), the State’s Children with Special Health Care Needs (CSHCN) Program. By law, all hospitals are required to conduct hearing screens on all infants delivered under their care and to notify the IDPH Newborn Hearing Program of the test results on all infants. Hearing screening is completed utilizing a non-invasive technology that allows the detection of hearing loss within a few hours of birth.

According to current estimates, permanent congenital hearing loss of greater than 25 dB HL (hearing level) in the poorer ear is present in at least three infants per thousand (White, 1997). Given Illinois birth rates, it would be expected that approximately 540 infants would be born each year with significant, congenital hearing loss. Research conducted by Yoshinaga-Itano (1995) indicates that children identified with hearing loss who are linked to appropriate

---

145 The Genetic Task Force of Illinois (GTFI) is Illinois’ primary professional organization for clinical genetic providers and has served in this capacity for 23 years. For more information, see http://www.gtfi.us
146 From conversation with IDPH, Genetics and Newborn Screening program staff, May 3, 2004.
147 Genetic and Metabolic Diseases Advisory Committee. Minutes—November 18, 2004 Meeting.
149 Information provided by the IDPH Newborn Hearing Program Staff.
intervention by 6 months of age can develop normal language skills. Early indications show that the state’s universal screening program is effective in identifying children with congenital hearing losses at an earlier age. Prior to universal newborn hearing screening implementation, a 1995 study by Kittrell and Arjmand revealed that in Illinois the average age of identification of hearing loss was 20.2 months and the average age of initial amplification was 31.7 months. By mid-calendar year 2005, two and a half years after universal newborn hearing screening implementation, the average age of diagnosis of infants reported to Public Health had greatly improved to 3.8 months of age.

Newborn Hearing Program staff has successfully trained all 137 birthing hospitals in Illinois to report their local newborn hearing screening data in an encrypted electronic format. Electronic reporting and data management is done using a central and 137 local applications of the Hi*Track Data Management System, the software system created by the National Center for Hearing Assessment and Management (NCHAM), Utah State University, for newborn hearing screening data management and tracking. Hospitals are required to send their screening data to IDPH weekly by encrypted email attachment. This allows for the timely submission of screening and follow-up data and a means of monitoring receipt of data by the State.

At the State level, Hi*Track is used for centralized, statewide program management of tracking and follow-up activities. Upon receipt of the data, the files are merged into Hi*Track from which letters to physicians and parents are generated, monitoring of tracking activities takes place, and data management, report generation and program evaluation can be done. Currently, there is not a protocol for the integration of newborn blood spot screening services with the Newborn Hearing Program and the program operates separately from the Genetics and Newborn Screening Program.

Illinois Regionalized Perinatal Program

The Perinatal Care Program is a comprehensive statewide system of inpatient services created to provide optimal care throughout pregnancy and early infancy in order to improve the health of women and infants. Services are provided to pregnant women who require high-risk delivery and newborn infants who require neonatal intensive care. The length of inpatient perinatal care varies based on the severity of illness and medical complications of the patients.

Four levels (capabilities) of perinatal care are well defined in administrative rules to provide a basis for quality assurance and accountability of hospital-based providers: basic or Level I, intermediate or Level II, specialty or Level II with extended capabilities, and sub-specialty or level III, with all facilities integrated into networks of care. Each region has a perinatal center that is required to establish and maintain a structure that ensures continuous quality improvement in perinatal care. This structure provides or coordinates an integrated program of quality improvement activities such as identification of important aspects of perinatal care (e.g., percentage of very low birth weight infants born in a Level III facility or Perinatal Center), collecting data on performance and taking necessary actions to solve problems or otherwise improve the quality and effectiveness of perinatal patient care and the regional system.
The IDPH currently operates a number of data systems to monitor and track the statewide incidence of genetic and metabolic conditions, the provision of follow up service to infants with positive newborn screens, and the activities of clinical genetics grantees. These include the following systems:

**Adverse Pregnancy Outcomes Reporting System**\(^{152}\) (APORS)

The 1984 Illinois Health and Hazardous Substances Registry Act (Public Act 83-1361) mandated the creation of three statewide registries to track the incidence of adverse pregnancy outcomes, cancer, and occupational disease. Representing one of these registries, the Adverse Pregnancy Outcomes Reporting System (APORS) tracks the incidence of all infants evidencing signs, symptoms, or pre-discharge diagnoses of: congenital anomaly, serious infection, endocrine, metabolic, and/or blood disorders, low birth weight (less than 1501 grams), or positive toxicities for controlled substances. In addition, APORS tracks the incidence of fetal and neonatal death as well as all infants with neonatal intensive care stays of more than 24 hours, whether or not they meet the additional APORS criteria.

Data are collected from neonatal intensive care unit logs and hospital discharge records, with hospital staff completing and submitting an infant discharge record to IDPH for every infant that the hospital determines meets criteria for inclusion in the APORS system. Hospitals are legally required to submit the form within seven days of an infant’s discharge. APORS also utilizes data provided by IDPH staff in response to certain APORS dispositions, and integrates information from fetal death certificates and birth certificates.

Designed primarily as a surveillance system, APORS also serves as the state’s referral system for high-risk infants. Hospitals submit completed infant discharge forms to the IDPH Division of Epidemiological Studies, APORS Program, as well as to the infant’s physician and the local health department serving the county in which the family resides. All APORS infants (up to 24 months of age) are eligible for follow up nursing services provided through the local health departments. These services are provided as a part of the High Risk Infant Follow-up Program, administered by the Illinois Department of Human Services (see below). Thus the APORS system serves as both a surveillance system, monitoring the occurrence of adverse pregnancy outcomes, many of which may result from genetic conditions, and a referral/service system, providing the information that initiates follow-up services through the local health departments.

The APORS system/High Risk Infant Follow-up Program\(^{153}\) is an important point of entry into genetic services in the local health department. Fifty-four percent of clients receiving genetic services in local health departments in 2004 were referred through these programs. APORS is, thus, programmatically linked to the Genetics and Newborn Screening Program, through services provided to APORS infants presumptive for, or diagnosed with, a genetic


\(^{153}\) It should be noted that at the local health department level, APORS and the High Risk Infant Follow-up Program are effectively the same program.
condition. Further, APORS follow-up service information is itself reported and maintained in the databases comprising the Genetic Newborn Screening System (see below), which are administered by the Genetics and Newborn Screening Program. Despite the programmatic linkage, however, there are no linkages between the data in these two data systems.

**Genetic Newborn Screening System (GNSS)**

Maintained by the Genetics and Newborn Screening Program, the GNSS contains newborn screening test and follow up information on each infant born in Illinois for whom a newborn screening specimen is received. Data maintained include: date of birth, sex, birth weight, gestational age, mother’s county of residence, hospital submitting the specimen, physician name, newborn screening test information, and follow up activities, including retests, referrals, diagnoses and treatment. For diagnosed cases, data are maintained on follow up and treatment activities and developmental progress through age 15, and includes information on the utilization of medical treatment products provided by IDPH to patients with PKU and certain other metabolic disorders. The GNSS also includes information related to sudden infant deaths (SIDS) that are reported to the IDPH SIDS/Infant Mortality Program. These SIDS records are directly linked to the newborn screening record. The GNSS also includes information utilized by the Perinatal Hepatitis B Prevention Program. The following programs contribute and/or utilize the information stored within this data system: the IDPH Genetics and Newborn Screening Program, the IDPH Newborn Screening Laboratory, the IDPH SIDS and Infant Mortality Program, and the IDPH Perinatal Hepatitis B Prevention Program.

**Electronic Birth Registry**

Maintained by the IDPH Division of Vital Records, the Birth Registry incorporates data from birth certificates on each infant born in Illinois. Data include, but are not limited to: date and place of birth, sex, race, ethnicity, name and age of mother/father, residence, birth weight, congenital malformations, pregnancy complications (including illnesses or other conditions affecting the pregnancy), and number of prenatal visits. Approximately 99 percent of this information is submitted to IDPH electronically, utilizing state distributed software, with paper copies of the birth certificate submitted by the hospital to the local county registrar, and then to IDPH.

**Death Registry**

Maintained by the Division of Vital Records, the death registry contains data on every individual that dies in the state of Illinois, including, but not limited to, information on all fetal deaths. Information includes: death by occurrence, cause of death, birth weight (for fetal deaths only), sex, race, location, and residence. For fetal deaths, information includes: number of prenatal visits and month prenatal care began, pregnancy complications, illness or conditions affecting pregnancy, and labor complications.
Genetic Counseling Services

Maintained by Genetics and Newborn Screening Program, the Genetic Counseling Services data file maintains patient information submitted by grant funded clinical genetic centers. IDPH patient data forms are completed by the clinical genetics grantees for each patient served and submitted to the Genetics and Newborn Screening Program quarterly. Data submitted include patient demographics, patient visit information (including source of referral and disposition), prenatal screening indicators, family history indicators, teratogen exposure, lab and other diagnostic tests utilized, clinical services rendered, and outcomes. It is important to note that centers differ in the comprehensiveness of the data they report, with some centers reporting data on all patients seen and others reporting data only for those patients that are seen through the grant program. Reports are generated annually detailing grantees activity and overall statewide activities and demographics of patients served.

Medical Services for Hemoglobinopathies\textsuperscript{154}

Maintained by the Genetics and Newborn Screening Program, this report maintains data on patients served by the pediatric hematologists funded through IDPH’s sickle cell grant program. Data include patient demographics, diagnosis and treatment, and are submitted quarterly to the Genetics and Newborn Screening Program by funded pediatric hematologists. Patient data include date of birth, race, referral source, screening results, final diagnosis, parent and sibling diagnosis, whether family received counseling, treatment start date, and source of payment.

Currently these data systems operate primarily on a mainframe system utilizing NOMAD\textsuperscript{\textregistered} software, and have only minimal linkage among their related data elements. An independent evaluation of the APORS surveillance system conducted in 2001 recommended, among other changes, a reorganization to include the systematic linkage of data elements relevant to maternal child health in the APORS, vital records, and GNSS registries, as well as a replacement of the mainframe system with a personal computer based system.\textsuperscript{155} Attempting to address this issue, the Genetics and Newborn Screening Program is working toward developing a Request for Proposal (RFP) for a new data system that will ideally enable integration and linkage across these and other data systems utilized in programs providing screening and clinical services to infants, children, and adults with genetic conditions and special health care needs.\textsuperscript{156}

While the above represent the primary data systems currently utilized by the programs supported by the Genetics and Newborn Screening Program, IDPH maintains other surveillance systems that may prove of relevance to genetics/genomics, especially as the role of genetics in chronic disease is increasingly understood. These include the:

\textsuperscript{154} Information provided by the IDPH staff.
\textsuperscript{156} Genetic and Metabolic Diseases Advisory Committee. Minutes—November 18, 2004 Meeting.
Pregnancy Risk Assessment Monitoring System (PRAMS)\textsuperscript{157}

The Illinois Pregnancy Risk Assessment Monitoring System (PRAMS) is an ongoing population-based survey of Illinois women who have delivered a live born infant in Illinois. PRAMS is an initiative by the U.S. Centers for Disease Control and Prevention (CDC) to reduce infant mortality and adverse birth outcomes. The unique information collected by the PRAMS project is used by health professionals, administrators, policy makers and researchers to develop and modify programs and policies to improve the health of women and children in Illinois. PRAMS collects information from mothers about their behaviors and experiences before, during and immediately following the birth of the baby. Data are collected on a variety of topics including: family planning, prenatal care, risk factors, and infant care and health.

Behavioral Risk Factor Surveillance System (BRFSS)

Administered by the Illinois Center for Health Statistics, the BRFSS is population-based data of behaviors and conditions among adults that are associated with health risks leading to premature death, morbidity, and other chronic diseases. Data are collected by telephone survey on Illinois residents eighteen years of age and older and include demographic information, health status and medical history information, as well as information related to physical activity, cancer screening (colorectal, mammography), weight control practices, smoking, alcohol consumption, health insurance, routine check ups and health care utilization, and nutrition.

Illinois State Cancer Registry

Maintained by the Division of Epidemiological Studies and authorized with APORS under the 1984 Illinois Health and Hazardous Substances Registry Act (Public Act 83-1361), the Cancer Registry maintains population based cancer incidence data submitted by hospitals, treatment centers, radiation therapy facilities, and pathology laboratories. Data collected include: patient demographics, tobacco/alcohol use, initial diagnosis date and method, stage of disease, treatment information, and survival status.

These three data files are maintained on a personal computer system.\textsuperscript{158}

Other Public Health Agencies

In administering the genetics program, IDPH coordinates with two other state agencies involved in the provision of services to individuals with genetic conditions and/or concerns: the Illinois Department of Human Services (IDHS) and the Division of Specialized Care for Children (DSCC).

\textsuperscript{157} Information provided by the IDPH staff.
\textsuperscript{158} See, IDPH. Office of Epidemiology and Health Systems Development. \textit{Database and Datafile Resource Guide}. 2002. \url{http://www.idph.state.il.us/pdf/ddrg02.PDF}.
Illinois Department of Human Services (IDHS)

The IDHS administers the Title V Maternal and Child Health Services Federal Block Grant through programs and services to pregnant women, infants, and children with various special health care needs, including genetic and metabolic conditions. Programs administered by IDHS that are currently most relevant to genetics include:

Special Supplemental Nutrition Program for Women, Infants, and Children Program (WIC). WIC provides nutrition education, supplemental foods, and referral to health care services to income eligible pregnant, breastfeeding and postpartum women and infants/children up to age five with the goals of reducing infant mortality and premature and low birth weight, and of facilitating the healthy growth and development of children.

Family Case Management (FCM). FCM provides support services and care coordination to income eligible families to ensure access to health care for pregnant women, infants, and young children. The program funds 116 agencies statewide, including local health departments, federally qualified health centers, and community based organizations. Case management services are also provided to infants up to 24 months of age identified through the APORS program. Children up to age two may also be case managed by FCM providers downstate if they meet the guidelines for high risk as defined in their local agency Policy and Procedure Manual. Some of the risk factors would be social, emotional, and environmental or health related to name a few.

Family Planning. The Family Planning program provides medical and counseling services, including pre-conceptual counseling, physical exams, laboratory tests and screenings, and risk assessment related to pregnancy, childbirth, and reproductive health.

High Risk Infant Follow-Up. The High Risk Infant Follow-up Program provides follow up clinical and support services through local health departments to infants up to 24 months of age with congenital infection, endocrine, metabolic, or immune disorder, blood disorder, low birth weight (less than 1501 grams), congenital anomaly or condition, and/or positive toxicity for controlled substance. The High Risk Infant Follow-Up Program is a component of the Family Case Management Program and is programmatically linked with the WIC, Perinatal Care, and Early Intervention Programs.

Pediatric Primary Care. Pediatric Primary Care provides primary care services through local health departments to infants and children. Services include immunizations, lead screening, hearing and vision screening, and other primary health care services.

Individuals with Disabilities Education Act, Part C Early Intervention Program. The Early Intervention Program is a statewide program of evaluation and assessment for infants and toddlers under three years of age and of services for those who have a disability, a 30 percent delay in development in any area, or are at risk of developmental delays. Services are provided to assist eligible children to develop basic developmental skills. Parents provide most of the care needed to help their children develop, guided by therapists who serve their children.

In addition, IDHS maintains the following database:
Family Case Management System (Cornerstone). Cornerstone is a centralized data management system developed by the state of Illinois and administered through the Illinois Department of Human Services to track information related to maternal and child health program client service utilization, clinical care, and outcomes. Cornerstone enables the maintenance of individual client files that are comprehensive for all maternal child health programs. In addition, Cornerstone may be queried to produce regional or statewide reports on service utilization and provision, among other things. Currently, Cornerstone is being transformed from a mainframe to an electronic system.

Programs that utilize the Cornerstone system include the following IDPH and IDHS programs: Family Case Management, Special Supplemental Nutrition Program for Women, Infants and Children (WIC), Immunization Program, Pediatric Primary Care, Early Intervention, Healthy Families Illinois, Illinois Diabetes Control Program, Healthy Start, and the Illinois Breast and Cervical Cancer Screening Program. Over 3.5 million individuals are tracked through the Cornerstone system. Local health departments utilize the Cornerstone system to maintain client data. Thus, infants and families provided with follow up services through the APORS or Genetics and Newborn Screening Program at local health departments may be included in the Cornerstone System. The Illinois Department of Public Health, however, currently does not have direct access to the Cornerstone System. Thus, for the IDPH Genetics and Newborn Screening Program, the local health departments serve as the primary point of access, when needed, to the Cornerstone System.

Division of Specialized Care for Children (DSCC)

DSCC is the Illinois Title V Program for Children with Special Health Care Needs. DSCC provides care coordination, benefits management and financial assistance for diagnostic and treatment services to children (through age 21) who meet financial eligibility and have, or are suspected of having, a treatable chronic medical condition in one of the following categories:

- Orthopedic conditions (bone, muscle, joint disease)
- Heart defects
- Hearing loss
- Neurological conditions (nerve, brain, spinal cord)
- Certain birth defects
- Disfiguring defects such as cleft lip, cleft palate, and severe burn scars
- Speech conditions which need medical/dental treatment
- Certain chronic disorders such as hemophilia and cystic fibrosis
- Certain inborn metabolic problems including phenylketonuria (PKU) and galactosemia
- Eye impairments including cataracts, glaucoma, strabismus and certain retinal conditions—excluding isolated refractive errors
- Urinary system impairments (kidney, ureter, bladder)

Through cooperative agreement with IDPH, DSCC covers the cost of diagnostic testing as needed for all infants with positive or suspect newborn screens. Children with conditions

---

159 Information on Cornerstone comes in part from the Illinois Primary Health Care Association, “Cornerstone General Information,” available online at http://www.cstonesupport.info/general/default.htm
and/or impairments that meet DSCC’s medical eligibility criteria may also receive continued services through DSCC based on financial eligibility. It is important to note that DSCC’s eligibility criteria are primarily impairment, not diagnosis, based. This means that although, for example, children with endocrine disorders or Down Syndrome (as opposed to cystic fibrosis, for example) may not be eligible for services on the basis of their diagnosis, they are eligible for services related to impairments falling within the categories of medical eligibility. Currently, impairments associated with gastrointestinal, hematology, endocrine, and pulmonology conditions are not medically eligible for service through DSCC.\textsuperscript{161} DSCC serves approximately 20,000 families from all of Illinois’ 102 counties annually through 13 regional offices\textsuperscript{162}.

**Clinical Genetics in Illinois**

Currently, over 20 organizations serve as primary providers of clinical genetic services in Illinois. These organizations include academic medical centers, private health systems/hospitals, a county hospital, and a state funded teratogen information service. Services are provided at multiple primary and outreach sites throughout the state, but remain concentrated in the Chicago metropolitan area (see Figure 3.2). Outside of the Chicago metropolitan area, clinical genetic centers are located in Rockford, Peoria, Urbana, Springfield, and St. Louis (serving Illinois residents in the southern part of the state). Key centers and services have been identified through interviews and secondary literature review. Centers that participate as grantees in IDPH’s clinical genetics program are identified by asterisk. \textit{It should be noted that this list may not be comprehensive.} Provision of the following list does not constitute in any way an endorsement of these centers or their services by the Illinois Department of Public Health or the University of Illinois at Chicago.

**Clinical Genetic Centers in the Chicago Metropolitan Area (in alphabetical order)**

1. \textit{Adventist Hinsdale Hospital} (Hinsdale)
2. \textit{Advocate Health Care*} (Oak Brook)
3. \textit{Children’s Memorial Hospital*} (Chicago)
4. \textit{Evanston Northwestern Healthcare} (Evanston)
5. \textit{Illinois Teratogen Information System*} (Chicago)
6. \textit{John H. Stroger Jr. Hospital of Cook County*} (Chicago)
7. \textit{Loyola University Medical Center*} (Maywood)
8. \textit{Northwestern University/Northwestern Memorial Hospital (NMH)/Northwestern Medical Faculty Foundation (NMFF)} (Chicago).
9. \textit{Reproductive Genetics Institute} (Chicago)
10. \textit{Rush University Medical Center*} (Chicago)
11. \textit{University of Chicago Hospitals*} (Chicago)
12. \textit{University of Illinois Medical Center at Chicago*} (Chicago)

**Clinical Genetic Centers outside of the Chicago metropolitan area (in order by location):**

13. \textit{Rockford Memorial Hospital*} (Rockford)
14. \textit{OSF St. Anthony Medical Center} (Rockford)
15. \textit{Southern Illinois University, School of Medicine*} (Springfield)

\textsuperscript{161} Information provided in interview.
\textsuperscript{162} To find an office, see http://internet.dscc.uic.edu/dsccroot/office_lookup.asp
Clinical Genetics Service Provision

As part of the interview process (see Appendix 1: Methods, and Chapter 6: Interview Findings), genetic service providers were asked to describe the process by which a typical patient receives genetic services, including how the patient is referred to genetic services, the process by which services are provided, their interaction with other medical specialists or health care providers, the issues they discuss with patients, and the types of referrals they make. They were also asked to describe the process of recommending or ordering genetic tests for patients. Following is a summary of clinical genetic service provision based on participant responses.

In terms of how patients and families obtain clinical genetic services in the current genetic services system, participants reported that most patients are referred to genetic specialists by their primary care provider, specialty physicians or from nurse midwives. Some patients are referred as inpatient consultations associated with the hospital, academic medical center or health care system, for example, a patient who is seeing a faculty obstetrician in an academic medical center, or a newborn in the nursery or neonatal care unit. Patients are also self-referred, requesting evaluation for themselves or their children, and are sometimes referred by family members. Other patients are referred from an in-house laboratory, when there is an abnormal genetic test result. Finally, some patients are referred from genetic coordinators at local health departments or through the state’s newborn screening program.

According to the genetic service providers interviewed, the process by which patients receive genetic evaluation and counseling varies by the type of genetic service or patients seen. For example, cancer genetic counseling is often more counselor-directed. The cancer genetics patients are often evaluated, counseled, offered genetic testing and follow up by a genetic counselor only, with the physician serving as a “back-up.” By contrast, in some prenatal settings, a genetic counselor works with a patient prior to the patient being seen by a maternal fetal medicine specialist, with a geneticist becoming involved only if the maternal fetal medicine physician feels that there is a need. Both a genetic counselor and a geneticist usually evaluate pediatric patients, with the genetic counselor obtaining family and medical histories and explaining inheritance and genetic testing, and the geneticist conducting the physical examination and evaluating the findings. Issues discussed with patients during genetic evaluation and counseling generally include: the actual or potential cause of the genetic condition; genetics (e.g. inheritance); genetic testing; management and intervention; possibility of risk to other family members; and ethical, legal and social issues, such as insurance and employment discrimination, when appropriate. The genetic counselors are frequently involved in coordinating the laboratory and other recommended testing, are responsible for writing the summary letter, and obtain and communicate laboratory results to the physician.

Genetic providers generally reported good working relationships with other specialists. Genetic providers indicated making a variety of referrals for follow-up evaluation and counseling, depending on the nature of the consultation. Prenatal referrals, for example, could include surgeons, neurologists, cardiologists, neonatologists and pulmonologists. Pediatric referrals include specialists such as cardiologists or neurologists, dentists, endocrinologists, and support groups – based on patient need. Referrals are also made to WIC for pre- and post-natal
patients. Cancer patients at risk for adverse psychological outcomes may be referred to psychologists. It was reported that in all genetic clinical settings (prenatal, pediatric, oncology), a summary letter is sent to the referring physician as communication of the genetic counseling and evaluation that has occurred.

Genetic service providers indicated that they consider a number of issues before recommending or ordering testing. The genetic history and evaluation frequently determine the recommendation for testing such as when there is a genetic abnormality or risk identified in a pregnancy; or when the results of genetic testing will change the management of a patient. The most significant issue reported is that of insurance. Genetic providers indicated that they consider the patient's insurance coverage because some insurance companies will not pay for genetic testing or only pay for certain genetic tests. For patients who receive public aid, this is a significant issue. As one genetic provider noted, we “want to give them the same standard of care…but we send a test or do a procedure and don’t get paid. The [Genetics] Department eats the cost.” Genetics providers indicated that there is a “two-tier” system in Illinois with regard to genetic testing. Those individuals/families that have insurance or money can get the genetic tests while those who have Medicaid or do not have insurance cannot. Genetic providers often need to get preauthorization to do genetic testing first and this can take time. As one provider noted, “Trying to get it (genetic testing) paid for is our big issue.” Other considerations on the part of genetics providers include whether the patient has the funds or wants to pay out of pocket.

Several factors that influence where a particular genetic test is sent were raised. Some institutions have their own laboratory (e.g. cytogenetics, DNA) that is used. In some institutions, the choice of laboratory is shaped by the institution’s contracts with specific laboratories. For example, several universities have contracts with Mayo Laboratories. The genetic testing sample is sent to Mayo and Mayo sends it to whomever they have a contract with to perform the test. Since some tests are only offered at one laboratory in the U.S. or internationally, genetic providers often use the GeneTests web site (www.genetests.org) to identify a laboratory where the specific genetic test is being performed. The quality of the laboratory, their detection rate and quality assurance are additional factors taken into consideration.

Genetic service providers reported several common issues discussed with patients regarding genetic testing as a part of the informed decision-making process. These include determining why a patient wants to pursue a genetic test; the risks, benefits and limitations of the genetic test; how the test will aid in a medical diagnosis and possible management or alter medical treatment; whether the testing is covered by insurance; and, for adult patients, potential for discrimination (insurance and employment) and the psychosocial aspects of predictive testing.

**Education and Research**

In addition to a public health and clinical genetics service infrastructure, Illinois has educational resources, including one active genetics residency program, sponsored jointly by the University of Chicago and Northwestern University, and one genetic counseling graduate program, which is located at Northwestern University. Researchers and practitioners affiliated with these and other universities and organizations are currently engaged in a broad range of clinical genetics research projects, addressing such issues as: the genetics of hypertension, pancreatic cancer, schizophrenia, breast and ovarian cancers, inflammatory bowel disease, and bipolar disorder. See Appendix 9 for a partial list of ongoing research projects in Illinois.
Chapter 4: Survey Findings: Local Health Departments

All 94 Illinois local health departments were surveyed by mail; details about the methodology are found in Appendix 1. The response rate for the local health department survey was 75.5% (71/94). The survey questionnaire is included in Appendix 2. Of the 71 respondents, 34 were from urban health departments and 37 were from rural health departments (based on the 2003 Rural-Urban Continuum Codes). Almost all respondents had a nursing degree. Of the 23 non-responding health departments, 6 were located in urban areas.

Fifty respondents (74%) indicated that their health department currently provides genetic services\textsuperscript{163}; of those, 39 (78%) received a grant from IDPH to provide genetic services. The following section refers to the 50 health departments that provide genetic services, with two subgroups based on whether they received funding from IDPH for genetic services (referred to as “funded” and “non-funded”).

Local Health Departments Providing Genetic Services

Provision of Genetic Services

Among funded health departments, almost all reported providing genetic screening and referrals to genetic providers (92% each; Table 4.1). (Note that genetic screening at a local health department involves not testing, but the use of a genetic screening tool to identify individuals at risk, or having a family history, of having or passing on a specific genetic disorder.) Most also noted their involvement in newborn screening follow-up and public education (74% each). Almost one-fourth of health departments had on-site genetic clinics. All non-funded health departments (n=11) provided newborn screening follow-up, and most were able to refer clients to a physician (83%) or genetic provider (73%). Only one non-funded respondent reported that the health department provided public or professional education on genetics.

Table 4.1 Genetic services provided by the health department

<table>
<thead>
<tr>
<th>Service</th>
<th>All health departments providing genetic services (n=50)</th>
<th>IDPH-grant Funded health departments (n=39)</th>
<th>Not funded health departments, providing genetic services (n=11)</th>
</tr>
</thead>
<tbody>
<tr>
<td>Newborn screening follow up</td>
<td>80%</td>
<td>74%</td>
<td>100%</td>
</tr>
<tr>
<td>Genetic screening*</td>
<td>78%</td>
<td>92%</td>
<td>27%</td>
</tr>
<tr>
<td>Onsite genetic clinic</td>
<td>18%</td>
<td>23%</td>
<td>0%</td>
</tr>
<tr>
<td>Referral to genetic provider</td>
<td>88%</td>
<td>92%</td>
<td>73%</td>
</tr>
<tr>
<td>Referral to physician</td>
<td>66%</td>
<td>61%</td>
<td>82%</td>
</tr>
<tr>
<td>Public education</td>
<td>60%</td>
<td>74%</td>
<td>9%</td>
</tr>
<tr>
<td>Professional education</td>
<td>42%</td>
<td>51%</td>
<td>9%</td>
</tr>
<tr>
<td>Sample collection</td>
<td>10%</td>
<td>13%</td>
<td>0%</td>
</tr>
<tr>
<td>Other</td>
<td>4%</td>
<td>5%</td>
<td>0%</td>
</tr>
</tbody>
</table>

\textsuperscript{163} The survey asked “Which of the following genetic services does your health department provide? “ and included a list of services, including an “other” category.
IDPH has a genetic screening tool that funded health departments are instructed to use (this tool is not a part of Cornerstone). Thirty-four (87%) grant funded health departments reported using this screening tool, along with one non-funded health department. Some funded (28%) and non-funded (36%) health departments reported using Cornerstone as their genetic screening tool, either alone or in addition to another screening tool (Table 4.2).

Table 4.2 Use of genetic screening tools

<table>
<thead>
<tr>
<th></th>
<th>All health departments providing genetic services (n=50)</th>
<th>IDPH-grant Funded health departments (n=39)</th>
<th>Not funded health departments, providing genetic services (n=11)</th>
</tr>
</thead>
<tbody>
<tr>
<td>IDPH</td>
<td>70%</td>
<td>87%</td>
<td>9%</td>
</tr>
<tr>
<td>Cornerstone</td>
<td>30%</td>
<td>28%</td>
<td>36%</td>
</tr>
<tr>
<td>Other</td>
<td>4%</td>
<td>5%</td>
<td>0%</td>
</tr>
</tbody>
</table>

Totals may equal more than 100%.

Most funded health departments, and one non-funded health department, advertised their genetic services (Table 4.3). Of the funded health departments that advertised, almost all (97%) did so to the general public and 74% did to physicians.

Table 4.3 Advertisement of genetic services

<table>
<thead>
<tr>
<th></th>
<th>All health departments providing genetic services (n=50)</th>
<th>IDPH-grant Funded health departments (n=39)</th>
<th>Not funded health departments, providing genetic services (n=11)</th>
</tr>
</thead>
<tbody>
<tr>
<td>Yes</td>
<td>64%</td>
<td>79%</td>
<td>9%(1)</td>
</tr>
<tr>
<td>No</td>
<td>36%</td>
<td>21%</td>
<td>91%</td>
</tr>
</tbody>
</table>

If yes, to whom were these services advertised:

<table>
<thead>
<tr>
<th></th>
<th>All health departments providing genetic services (n=50)</th>
<th>IDPH-grant Funded health departments (n=39)</th>
<th>Not funded health departments, providing genetic services (n=11)</th>
</tr>
</thead>
<tbody>
<tr>
<td>Physicians</td>
<td>75%</td>
<td>74%</td>
<td>9%</td>
</tr>
<tr>
<td>Nurse Practitioners</td>
<td>47%</td>
<td>45%</td>
<td>9%</td>
</tr>
<tr>
<td>Hospitals</td>
<td>50%</td>
<td>48%</td>
<td>9%</td>
</tr>
<tr>
<td>Birthing Centers</td>
<td>13%</td>
<td>10%</td>
<td>9%</td>
</tr>
<tr>
<td>Consumer Groups</td>
<td>22%</td>
<td>23%</td>
<td>0%</td>
</tr>
<tr>
<td>General Public</td>
<td>97%</td>
<td>97%</td>
<td>9%</td>
</tr>
<tr>
<td>Other Health Departments</td>
<td>28%</td>
<td>29%</td>
<td>0%</td>
</tr>
<tr>
<td>Community Health Centers</td>
<td>34%</td>
<td>35%</td>
<td>0%</td>
</tr>
<tr>
<td>Genetic Centers</td>
<td>6%</td>
<td>3%</td>
<td>9%</td>
</tr>
<tr>
<td>Other</td>
<td>6%</td>
<td>6%</td>
<td>0%</td>
</tr>
</tbody>
</table>
The most common health department programs that were reported to have involvement with the provision of genetic services were Family Case Management (96%), APORS (94%), WIC (78%), and Teen Parent Services (56%). Few health departments reported involvement by other program areas, such as diabetes, osteoporosis, cardiovascular disease, and cancer, although almost half of non-funded health departments noted involvement by their Hearing/Vision Screening program (Table 4.4).

Table 4.4 Health department programs currently involved in the provision of genetic services

<table>
<thead>
<tr>
<th>Health Department Program Area</th>
<th>All health departments providing genetic services (n=50)</th>
<th>IDPH-grant Funded health departments (n=39)</th>
<th>Not funded health departments, providing genetic services (n=11)</th>
</tr>
</thead>
<tbody>
<tr>
<td>Genetics</td>
<td>66%</td>
<td>81%</td>
<td>9%</td>
</tr>
<tr>
<td>APORS</td>
<td>94%</td>
<td>95%</td>
<td>91%</td>
</tr>
<tr>
<td>WIC</td>
<td>78%</td>
<td>81%</td>
<td>64%</td>
</tr>
<tr>
<td>Family Case Mgt.</td>
<td>96%</td>
<td>97%</td>
<td>91%</td>
</tr>
<tr>
<td>Family Planning</td>
<td>38%</td>
<td>36%</td>
<td>46%</td>
</tr>
<tr>
<td>Teen Parent Services</td>
<td>56%</td>
<td>59%</td>
<td>46%</td>
</tr>
<tr>
<td>Doula</td>
<td>4%</td>
<td>5%</td>
<td>0%</td>
</tr>
<tr>
<td>Perinatal Care</td>
<td>10%</td>
<td>8%</td>
<td>18%</td>
</tr>
<tr>
<td>AOK, 0-3</td>
<td>6%</td>
<td>5%</td>
<td>9%</td>
</tr>
<tr>
<td>Hearing/Vision Screening</td>
<td>16%</td>
<td>8%</td>
<td>46%</td>
</tr>
<tr>
<td>Diabetes</td>
<td>10%</td>
<td>5%</td>
<td>27%</td>
</tr>
<tr>
<td>Osteoporosis</td>
<td>4%</td>
<td>0%</td>
<td>18%</td>
</tr>
<tr>
<td>Cardiovascular disease</td>
<td>2%</td>
<td>0%</td>
<td>9%</td>
</tr>
<tr>
<td>Cancer</td>
<td>6%</td>
<td>3%</td>
<td>18%</td>
</tr>
<tr>
<td>Other</td>
<td>12%</td>
<td>15%</td>
<td>0%</td>
</tr>
</tbody>
</table>

Staffing for Genetic Services Provision

None of the non-funded health departments reported having a genetic coordinator; 24 funded health departments had at least one person in that role. Funded health departments also reported utilizing nurses (29 health departments) to provide genetic services, as well as case managers (6), social workers (3), or other personnel (4). Non-funded health departments utilized nurses (9 health departments), case managers (5), and social workers or others (1 each) to deliver genetic services.
Meeting Clients’ Needs

Two-thirds of health departments felt that their current provision of genetic services adequately met the needs of their clients (Table 4.5). More funded (74%) than non-funded (45%) health departments reported being able to meet these needs. Reasons for not currently meeting needs included staffing constraints (e.g., unable to provide education to all people screened who have positive indicators, unable to complete screening tool on everyone who could be screened; unable to provide enough educational outreach opportunities as well as training for other health departments in region) as well as client access and follow-up problems (e.g., genetic counseling services are 1.5 hours away; many clients do not follow-up —“ I think our staff needs more education in presenting information to the client”).

Table 4.5 Health departments’ current provision of genetic services adequately met clients’ needs

<table>
<thead>
<tr>
<th></th>
<th>All health departments providing genetic services (n=50)</th>
<th>IDPH-grant Funded health departments (n=39)</th>
<th>Not funded health departments, providing genetic services (n=11)</th>
</tr>
</thead>
<tbody>
<tr>
<td>Yes</td>
<td>67%</td>
<td>74%</td>
<td>45%</td>
</tr>
<tr>
<td>No</td>
<td>20%</td>
<td>21%</td>
<td>18%</td>
</tr>
<tr>
<td>Don’t Know</td>
<td>12%</td>
<td>5%</td>
<td>36%</td>
</tr>
</tbody>
</table>

All Respondents

This section includes all health department respondents. Here, the two subgroups are (1) IDPH-funded health departments, and (2) health departments who do not receive funding, regardless of whether or not they provided genetic services. Respondents were asked about their perception of the impact of advances in human genetics on their health department programs and which programs will need to incorporate genetics information in the next three to five years. They were also asked a number of questions about the role of their health departments in providing different genetic services and their comfort level in doing so. They were also asked to discuss access barriers, unmet patient needs, major issues facing genetic service provision, and how IDPH could better support their efforts.

Future Program Needs

Funded health departments anticipated that recent advances in genetics would have a greater impact on their health departments programs in the next 5-10 and 10-15 years than did health departments without IDPH-funding (Table 4.6). When asked which chronic disease programs would need to incorporate genetic information in the next 3-5 years (Table 4.7), the most common responses were diabetes (62% of health departments; 51% of funded and 75% of not funded), cardiovascular (61%, 67%, and 53%), obesity (55%, 61%, and 47%), and cancer (51%, 54%, and 47%). About one-third and one-fourth of health departments saw a need for
asthma and Alzheimer’s programs, respectively, to incorporate genetic information. There was uncertainty as to whether there are other public health program areas that will have to address genetic needs in the future (Table 4.8).

Table 4.6 Anticipated impact of advances in genetics on health department programs.

<table>
<thead>
<tr>
<th></th>
<th>None</th>
<th>Very Little</th>
<th>Some</th>
<th>A lot</th>
<th>Don’t Know</th>
</tr>
</thead>
<tbody>
<tr>
<td>Currently</td>
<td>9 (14%)</td>
<td>21 (33%)</td>
<td>25 (40%)</td>
<td>3 (5%)</td>
<td>5 (8%)</td>
</tr>
<tr>
<td>In the next 5-10 years</td>
<td>3 (5%)</td>
<td>6 (9%)</td>
<td>29 (43%)</td>
<td>18 (27%)</td>
<td>11 (16%)</td>
</tr>
<tr>
<td>In the next 10-15 years</td>
<td>3 (4%)</td>
<td>5 (7%)</td>
<td>15 (22%)</td>
<td>33 (48%)</td>
<td>13 (19%)</td>
</tr>
</tbody>
</table>

<table>
<thead>
<tr>
<th></th>
<th>None</th>
<th>Very Little</th>
<th>Some</th>
<th>A lot</th>
<th>Don’t Know</th>
</tr>
</thead>
<tbody>
<tr>
<td>IDPH grant funded health departments (n=39)</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Currently</td>
<td>5 (15%)</td>
<td>10 (29%)</td>
<td>15 (44%)</td>
<td>2 (6%)</td>
<td>2 (6%)</td>
</tr>
<tr>
<td>In the next 5-10 years</td>
<td>1 (3%)</td>
<td>3 (8%)</td>
<td>17 (45%)</td>
<td>14 (37%)</td>
<td>3 (8%)</td>
</tr>
<tr>
<td>In the next 10-15 years</td>
<td>1 (3%)</td>
<td>2 (5%)</td>
<td>6 (16%)</td>
<td>24 (63%)</td>
<td>5 (13%)</td>
</tr>
</tbody>
</table>

<table>
<thead>
<tr>
<th></th>
<th>None</th>
<th>Very Little</th>
<th>Some</th>
<th>A lot</th>
<th>Don’t Know</th>
</tr>
</thead>
<tbody>
<tr>
<td>Not funded health departments (n=32)</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Currently</td>
<td>4 (14%)</td>
<td>11 (38%)</td>
<td>10 (35%)</td>
<td>1 (3%)</td>
<td>3 (10%)</td>
</tr>
<tr>
<td>In the next 5-10 years</td>
<td>2 (7%)</td>
<td>3 (10%)</td>
<td>12 (41%)</td>
<td>4 (14%)</td>
<td>8 (28%)</td>
</tr>
<tr>
<td>In the next 10-15 years</td>
<td>2 (7%)</td>
<td>3 (10%)</td>
<td>9 (29%)</td>
<td>9 (29%)</td>
<td>8 (26%)</td>
</tr>
</tbody>
</table>
Table 4.7 Chronic disease programs that will need to incorporate genetics information over the next 3-5 years.

<table>
<thead>
<tr>
<th></th>
<th>All health departments providing genetic services (n=71)</th>
<th>IDPH-grant Funded health departments (n=39)</th>
<th>Not funded health departments, (n=32)</th>
</tr>
</thead>
<tbody>
<tr>
<td>Asthma</td>
<td>32%</td>
<td>31%</td>
<td>34%</td>
</tr>
<tr>
<td>Cancer</td>
<td>51%</td>
<td>54%</td>
<td>47%</td>
</tr>
<tr>
<td>Cardiovascular</td>
<td>61%</td>
<td>67%</td>
<td>53%</td>
</tr>
<tr>
<td>Diabetes</td>
<td>62%</td>
<td>51%</td>
<td>75%</td>
</tr>
<tr>
<td>Alzheimer’s</td>
<td>25%</td>
<td>26%</td>
<td>25%</td>
</tr>
<tr>
<td>Obesity</td>
<td>55%</td>
<td>61%</td>
<td>47%</td>
</tr>
<tr>
<td>Osteoporosis</td>
<td>3%</td>
<td>3%</td>
<td>3%</td>
</tr>
<tr>
<td>Arthritis</td>
<td>7%</td>
<td>8%</td>
<td>6%</td>
</tr>
<tr>
<td>Other (1-Autism, 2-Mental Health)</td>
<td>4%</td>
<td>5%</td>
<td>3%</td>
</tr>
</tbody>
</table>

Table 4.8 Need for other public health program areas to incorporate genetics information over the next 3-5 years.

<table>
<thead>
<tr>
<th></th>
<th>All health departments providing genetic services (n=71)</th>
<th>IDPH-grant Funded health departments (n=39)</th>
<th>Not funded health departments, (n=32)</th>
</tr>
</thead>
<tbody>
<tr>
<td>Yes</td>
<td>24%</td>
<td>28%</td>
<td>19%</td>
</tr>
<tr>
<td>No</td>
<td>21%</td>
<td>14%</td>
<td>30%</td>
</tr>
<tr>
<td>Don’t Know</td>
<td>56%</td>
<td>58%</td>
<td>52%</td>
</tr>
</tbody>
</table>

The Health Department’s Role and Comfort in Providing Genetic Services

We asked respondents to indicate the importance of their health department’s role in providing a number of genetics or genetics-related services, as well as their comfort level regarding the ability of their health department to provide these services (Tables 4.9-4.11). Note that some of the services on the list may only be appropriate for certain types of health professionals to provide. Respondents who indicated “not applicable” were excluded from the totals. Note, however, that there are a few services for which a substantial number of respondents chose “not applicable;” there are different possible interpretations about why not applicable was chosen.

The importance of the health department role in most of the services included in the survey is reflected in the large percentage of respondents who rated the items as “very important” or “important.” For example, 98% reported that the health department’s role was very important or important in providing public/patient education about birth defect prevention strategies, and 97% reported the same for follow-up for positive newborn screening tests and for client referral to birth defect/genetic disease support services and programs. Less importance was assigned to a variety of other activities included in the survey. The trends were similar for
funded versus non-funded health departments, with non-funded health departments giving lower ratings than funded health departments.

The percentage of respondents who felt very comfortable or comfortable with their health department providing these services was much lower than the percentage that felt the services were very important or important. Funded health departments reported more comfort with all but one item than those without IDPH funding. Even the funded health departments indicated a low comfort level with several services that were rated as very important or important such as providing public/patient education about: “inheritance patterns and principles” (76% very important or important, 23% very comfortable or comfortable); “genetic risk in common chronic diseases” (89%, 32%); “genetic risk related to gene-environment interaction” (72%, 20%); and “social and legal issues related to genetic testing” (74%, 9%). The health departments without funding reported only a 50% comfort level with such as providing public/patient education about “birth defect prevention strategies” and providing patient services for “follow-up for positive newborn screening tests.”
Table 4.9 All respondents - How important do you think the following services are to your health department’s role and what is your level of comfort with the ability of your health department to provide each service?

<table>
<thead>
<tr>
<th>Service</th>
<th>Very Important</th>
<th>Important</th>
<th>Somewhat Important</th>
<th>Not Important</th>
<th>Not Applicable</th>
<th>Very Comfortable</th>
<th>Comfortable</th>
<th>Somewhat Comfortable</th>
<th>Not Comfortable</th>
<th>Not Applicable</th>
</tr>
</thead>
<tbody>
<tr>
<td>a. Public/Patient Education About:</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Inheritance patterns and principles</td>
<td>32%</td>
<td>44%</td>
<td>20%</td>
<td>3%</td>
<td>7</td>
<td>5%</td>
<td>14%</td>
<td>49%</td>
<td>37%</td>
<td>8</td>
</tr>
<tr>
<td>Birth defect prevention strategies</td>
<td>70%</td>
<td>28%</td>
<td>2%</td>
<td>0%</td>
<td>4</td>
<td>24%</td>
<td>38%</td>
<td>24%</td>
<td>14%</td>
<td>4</td>
</tr>
<tr>
<td>Possible genetic risks to offspring</td>
<td>48%</td>
<td>41%</td>
<td>10%</td>
<td>2%</td>
<td>5</td>
<td>7%</td>
<td>23%</td>
<td>42%</td>
<td>29%</td>
<td>5</td>
</tr>
<tr>
<td>Genetic risk in common chronic diseases</td>
<td>37%</td>
<td>47%</td>
<td>16%</td>
<td>0%</td>
<td>5</td>
<td>2%</td>
<td>22%</td>
<td>34%</td>
<td>42%</td>
<td>5</td>
</tr>
<tr>
<td>Genetic risk related to gene-environment interactions</td>
<td>32%</td>
<td>37%</td>
<td>32%</td>
<td>0%</td>
<td>7</td>
<td>0%</td>
<td>15%</td>
<td>19%</td>
<td>66%</td>
<td>7</td>
</tr>
<tr>
<td>Options for genetic testing</td>
<td>44%</td>
<td>43%</td>
<td>10%</td>
<td>3%</td>
<td>6</td>
<td>3%</td>
<td>30%</td>
<td>30%</td>
<td>30%</td>
<td>37%</td>
</tr>
<tr>
<td>Social and legal issues related to genetic testing (e.g., insurance discrimination)</td>
<td>30%</td>
<td>40%</td>
<td>25%</td>
<td>5%</td>
<td>7</td>
<td>0%</td>
<td>5%</td>
<td>31%</td>
<td>64%</td>
<td>7</td>
</tr>
<tr>
<td>Social and legal issues related to genetic testing (e.g., insurance discrimination)</td>
<td>30%</td>
<td>40%</td>
<td>25%</td>
<td>5%</td>
<td>7</td>
<td>0%</td>
<td>5%</td>
<td>31%</td>
<td>64%</td>
<td>7</td>
</tr>
<tr>
<td>b. Patient Services</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Identify genetic risk factors for disease.</td>
<td>44%</td>
<td>42%</td>
<td>15%</td>
<td>0%</td>
<td>6</td>
<td>9%</td>
<td>30%</td>
<td>42%</td>
<td>19%</td>
<td>7</td>
</tr>
<tr>
<td>Counsel patients regarding genetic testing options</td>
<td>41%</td>
<td>45%</td>
<td>13%</td>
<td>2%</td>
<td>12</td>
<td>8%</td>
<td>21%</td>
<td>36%</td>
<td>36%</td>
<td>12</td>
</tr>
<tr>
<td>Order genetic screening tests</td>
<td>32%</td>
<td>38%</td>
<td>13%</td>
<td>17%</td>
<td>20</td>
<td>3%</td>
<td>10%</td>
<td>5%</td>
<td>82%</td>
<td>24</td>
</tr>
<tr>
<td>Interpret results of genetic tests for clients</td>
<td>32%</td>
<td>36%</td>
<td>21%</td>
<td>11%</td>
<td>23</td>
<td>0%</td>
<td>13%</td>
<td>3%</td>
<td>85%</td>
<td>24</td>
</tr>
<tr>
<td>Follow-up for positive newborn screening tests</td>
<td>66%</td>
<td>31%</td>
<td>3%</td>
<td>0%</td>
<td>5</td>
<td>24%</td>
<td>38%</td>
<td>22%</td>
<td>16%</td>
<td>5</td>
</tr>
<tr>
<td>Care coordination for genetic diseases/birth defects</td>
<td>64%</td>
<td>26%</td>
<td>9%</td>
<td>2%</td>
<td>11</td>
<td>19%</td>
<td>28%</td>
<td>26%</td>
<td>26%</td>
<td>9</td>
</tr>
<tr>
<td>Medical management for genetic diseases/birth defects</td>
<td>50%</td>
<td>33%</td>
<td>12%</td>
<td>6%</td>
<td>14</td>
<td>7%</td>
<td>22%</td>
<td>27%</td>
<td>44%</td>
<td>18</td>
</tr>
<tr>
<td>Client referral to birth defect/ genetic disease support services and programs</td>
<td>62%</td>
<td>35%</td>
<td>3%</td>
<td>0%</td>
<td>5</td>
<td>17%</td>
<td>47%</td>
<td>24%</td>
<td>12%</td>
<td>5</td>
</tr>
<tr>
<td>Communicate with insurance companies to obtain coverage for genetic services for clients</td>
<td>34%</td>
<td>27%</td>
<td>27%</td>
<td>11%</td>
<td>23</td>
<td>0%</td>
<td>3%</td>
<td>27%</td>
<td>70%</td>
<td>26</td>
</tr>
</tbody>
</table>
**Table 4.10 Funded - How important do you think the following services are to your health department’s role and what is your level of comfort with the ability of your health department to provide each service?**

<table>
<thead>
<tr>
<th>Service Description</th>
<th>Very Important</th>
<th>Important</th>
<th>Somewhat Important</th>
<th>Not Important</th>
<th>Not Applicable</th>
<th>Very Comfortable</th>
<th>Comfortable</th>
<th>Somewhat Comfortable</th>
<th>Not Comfortable</th>
<th>Not Applicable</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>a. Public/Patient Education About:</strong></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Inheritance patterns and principles</td>
<td>38%</td>
<td>38%</td>
<td>22%</td>
<td>3%</td>
<td>0</td>
<td>6%</td>
<td>17%</td>
<td>50%</td>
<td>28%</td>
<td>1</td>
</tr>
<tr>
<td>Birth defect prevention strategies</td>
<td>76%</td>
<td>24%</td>
<td>0%</td>
<td>0%</td>
<td>0</td>
<td>30%</td>
<td>41%</td>
<td>19%</td>
<td>11%</td>
<td>1</td>
</tr>
<tr>
<td>Possible genetic risks to offspring</td>
<td>58%</td>
<td>34%</td>
<td>5%</td>
<td>3%</td>
<td>0</td>
<td>5%</td>
<td>32%</td>
<td>41%</td>
<td>22%</td>
<td>1</td>
</tr>
<tr>
<td>Genetic risk in common chronic diseases</td>
<td>43%</td>
<td>46%</td>
<td>11%</td>
<td>0%</td>
<td>0</td>
<td>3%</td>
<td>29%</td>
<td>26%</td>
<td>41%</td>
<td>1</td>
</tr>
<tr>
<td>Genetic risk related to gene-environment interactions</td>
<td>39%</td>
<td>33%</td>
<td>28%</td>
<td>0%</td>
<td>1</td>
<td>0%</td>
<td>20%</td>
<td>14%</td>
<td>66%</td>
<td>2</td>
</tr>
<tr>
<td>Options for genetic testing</td>
<td>50%</td>
<td>39%</td>
<td>8%</td>
<td>3%</td>
<td>1</td>
<td>3%</td>
<td>39%</td>
<td>25%</td>
<td>33%</td>
<td>2</td>
</tr>
<tr>
<td>Social and legal issues related to genetic testing (e.g., insurance discrimination)</td>
<td>37%</td>
<td>37%</td>
<td>17%</td>
<td>9%</td>
<td>2</td>
<td>0%</td>
<td>9%</td>
<td>35%</td>
<td>56%</td>
<td>3</td>
</tr>
<tr>
<td><strong>b. Patient Services</strong></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Identify genetic risk factors for disease</td>
<td>54%</td>
<td>38%</td>
<td>8%</td>
<td>0%</td>
<td>1</td>
<td>12%</td>
<td>39%</td>
<td>36%</td>
<td>12%</td>
<td>3</td>
</tr>
<tr>
<td>Counsel patients regarding genetic testing options</td>
<td>47%</td>
<td>35%</td>
<td>15%</td>
<td>3%</td>
<td>4</td>
<td>10%</td>
<td>26%</td>
<td>29%</td>
<td>35%</td>
<td>6</td>
</tr>
<tr>
<td>Order genetic screening tests</td>
<td>39%</td>
<td>39%</td>
<td>4%</td>
<td>18%</td>
<td>10</td>
<td>4%</td>
<td>13%</td>
<td>9%</td>
<td>74%</td>
<td>13</td>
</tr>
<tr>
<td>Interpret results of genetic tests for clients</td>
<td>46%</td>
<td>38%</td>
<td>4%</td>
<td>13%</td>
<td>14</td>
<td>0%</td>
<td>19%</td>
<td>5%</td>
<td>76%</td>
<td>15</td>
</tr>
<tr>
<td>Follow-up for positive newborn screening tests</td>
<td>73%</td>
<td>27%</td>
<td>0%</td>
<td>0%</td>
<td>0</td>
<td>32%</td>
<td>38%</td>
<td>21%</td>
<td>9%</td>
<td>1</td>
</tr>
<tr>
<td>Care coordination for genetic diseases/birth defects</td>
<td>76%</td>
<td>21%</td>
<td>0%</td>
<td>3%</td>
<td>7</td>
<td>29%</td>
<td>32%</td>
<td>14%</td>
<td>25%</td>
<td>6</td>
</tr>
<tr>
<td>Medical management for genetic diseases/birth defects</td>
<td>63%</td>
<td>30%</td>
<td>0%</td>
<td>7%</td>
<td>7</td>
<td>12%</td>
<td>24%</td>
<td>28%</td>
<td>36%</td>
<td>10</td>
</tr>
<tr>
<td>Client referral to birth defect/ genetic disease support services and programs</td>
<td>62%</td>
<td>35%</td>
<td>3%</td>
<td>0%</td>
<td>1</td>
<td>21%</td>
<td>52%</td>
<td>21%</td>
<td>6%</td>
<td>2</td>
</tr>
<tr>
<td>Communicate with insurance companies to obtain coverage for genetic services for clients</td>
<td>40%</td>
<td>32%</td>
<td>16%</td>
<td>12%</td>
<td>12</td>
<td>0%</td>
<td>0%</td>
<td>30%</td>
<td>70%</td>
<td>15</td>
</tr>
</tbody>
</table>
Table 4.11 Not funded - How important do you think the following services are to your health department’s role and what is your level of comfort with the ability of your health department to provide each service?

<table>
<thead>
<tr>
<th>Service</th>
<th>Very Important</th>
<th>Important</th>
<th>Somewhat Important</th>
<th>Not Important</th>
<th>Not Applicable</th>
<th>Very Comfortable</th>
<th>Comfortable</th>
<th>Somewhat Comfortable</th>
<th>Not Comfortable</th>
<th>Not Applicable</th>
</tr>
</thead>
<tbody>
<tr>
<td>a. Public/Patient Education About:</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Inheritance patterns and principles</td>
<td>23%</td>
<td>55%</td>
<td>18%</td>
<td>5%</td>
<td>7</td>
<td>5%</td>
<td>10%</td>
<td>48%</td>
<td>38%</td>
<td>7</td>
</tr>
<tr>
<td>Birth defect prevention strategies</td>
<td>62%</td>
<td>35%</td>
<td>4%</td>
<td>0%</td>
<td>4</td>
<td>15%</td>
<td>35%</td>
<td>31%</td>
<td>19%</td>
<td>3</td>
</tr>
<tr>
<td>Possible genetic risks to offspring</td>
<td>32%</td>
<td>52%</td>
<td>16%</td>
<td>0%</td>
<td>5</td>
<td>8%</td>
<td>8%</td>
<td>44%</td>
<td>40%</td>
<td>4</td>
</tr>
<tr>
<td>Genetic risk in common chronic diseases</td>
<td>28%</td>
<td>48%</td>
<td>24%</td>
<td>0%</td>
<td>5</td>
<td>0%</td>
<td>12%</td>
<td>44%</td>
<td>44%</td>
<td>4</td>
</tr>
<tr>
<td>Genetic risk related to gene-environment interactions</td>
<td>21%</td>
<td>42%</td>
<td>38%</td>
<td>0%</td>
<td>6</td>
<td>0%</td>
<td>8%</td>
<td>25%</td>
<td>67%</td>
<td>5</td>
</tr>
<tr>
<td>Options for genetic testing</td>
<td>36%</td>
<td>48%</td>
<td>12%</td>
<td>4%</td>
<td>5</td>
<td>4%</td>
<td>17%</td>
<td>38%</td>
<td>42%</td>
<td>4</td>
</tr>
<tr>
<td>Social and legal issues related to genetic testing (e.g., insurance discrimination)</td>
<td>20%</td>
<td>44%</td>
<td>36%</td>
<td>0%</td>
<td>5</td>
<td>0%</td>
<td>0%</td>
<td>24%</td>
<td>76%</td>
<td>4</td>
</tr>
<tr>
<td>b. Patient Services</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Identify genetic risk factors for disease</td>
<td>28%</td>
<td>48%</td>
<td>24%</td>
<td>0%</td>
<td>5</td>
<td>4%</td>
<td>17%</td>
<td>50%</td>
<td>29%</td>
<td>4</td>
</tr>
<tr>
<td>Counsel patients regarding genetic testing options</td>
<td>32%</td>
<td>59%</td>
<td>9%</td>
<td>0%</td>
<td>8</td>
<td>5%</td>
<td>14%</td>
<td>45%</td>
<td>36%</td>
<td>6</td>
</tr>
<tr>
<td>Order genetic screening tests</td>
<td>21%</td>
<td>37%</td>
<td>26%</td>
<td>16%</td>
<td>10</td>
<td>0%</td>
<td>6%</td>
<td>0%</td>
<td>94%</td>
<td>11</td>
</tr>
<tr>
<td>Interpret results of genetic tests for clients</td>
<td>15%</td>
<td>35%</td>
<td>40%</td>
<td>10%</td>
<td>9</td>
<td>0%</td>
<td>5%</td>
<td>0%</td>
<td>95%</td>
<td>9</td>
</tr>
<tr>
<td>Follow-up for positive newborn screening tests</td>
<td>56%</td>
<td>36%</td>
<td>8%</td>
<td>0%</td>
<td>5</td>
<td>13%</td>
<td>38%</td>
<td>25%</td>
<td>25%</td>
<td>4</td>
</tr>
<tr>
<td>Care coordination for genetic diseases/birth defects</td>
<td>50%</td>
<td>31%</td>
<td>19%</td>
<td>0%</td>
<td>4</td>
<td>8%</td>
<td>24%</td>
<td>40%</td>
<td>28%</td>
<td>3</td>
</tr>
<tr>
<td>Medical management for genetic diseases/birth defects</td>
<td>32%</td>
<td>36%</td>
<td>27%</td>
<td>5%</td>
<td>7</td>
<td>0%</td>
<td>20%</td>
<td>25%</td>
<td>55%</td>
<td>8</td>
</tr>
<tr>
<td>Client referral to birth defect/ genetic disease support services and programs</td>
<td>62%</td>
<td>35%</td>
<td>4%</td>
<td>0%</td>
<td>4</td>
<td>12%</td>
<td>40%</td>
<td>28%</td>
<td>20%</td>
<td>3</td>
</tr>
<tr>
<td>Communicate with insurance companies to obtain coverage for genetic services for clients</td>
<td>26%</td>
<td>21%</td>
<td>42%</td>
<td>11%</td>
<td>11</td>
<td>0%</td>
<td>6%</td>
<td>24%</td>
<td>71%</td>
<td>11</td>
</tr>
</tbody>
</table>
Barriers and Areas of Unmet Need

Respondents identified a number of barriers to patient access and/or areas of unmet need with respect to medical genetic services in Illinois. A frequently reported barrier was client/public knowledge, awareness, interest, and education about what genetics is and how genetic services can benefit them. Respondents reported frustration with getting clients of low education and socioeconomic status to appreciate the importance of genetic counseling and other services, and to follow-up on appointments and referrals. However, even clients with higher education often could not afford to follow up on referrals due to lack of insurance coverage for genetic services.

Other barriers and areas of unmet need cited included:
- Limited access — including lack of providers and lack of referral resources - to local genetic services, particularly in rural areas;
- Lack of transportation;
- Cost concerns — lack of insurance, insured but no coverage for genetic services, unable to pay out-of-pocket, Medicaid coverage only, concerns about what will happen to insurance coverage if a genetic condition is discovered;
- Limited genetic knowledge of local physicians and nurses, including where and how to refer patients for genetics services;
- Fears — of the unknown, in general, or of having a child or family member “labeled;” and
- Language — trying to find genetic providers who are bilingual, or have translators.

Key Issues Facing Local Health Departments

We also asked this question: In your opinion, what are the biggest issues public health care providers in local health departments will face over the next 5 to 10 years with respect to the provision of genetic services in Illinois? The response below summarizes much of what we heard from respondents:

“Educating ourselves and our community members that genetics is not just something happening in a lab or in research. Genetics is everywhere. So many chronic illnesses have a genetic link — this is our opportunity to educate!”

This quote reflects concerns about the issue of public education and the role of local health departments. Respondents recognized that as knowledge increases, so will the number of public inquiries about, for example, genetics and chronic conditions. They acknowledged that they will need to help clients understand which genetic services are available and why genetics is important.

The issue of increased need for public education is related to another set of concerns involving staffing. Respondents indicated the need for an adequate number of genetics staff, and for adequate training for nurses and health department staff in genetics (e.g., taking family histories, making referrals, understanding insurance coverage). Responses indicated that it is difficult for staff to keep up-to-date with available resources and new knowledge. A need for better coordination of care was also mentioned.
Other responses to this question included:

- Incorporating genetics into chronic disease programs – how to do counseling; how to provide services beyond using a genetic screening tool; responding to public requests for information;
- Funding – how to meet needs for direct services, research, staffing, etc. with limited funding and state budget constraints. “New initiatives, no new funding;”
- Problems with insurance coverage; fear of insurance discrimination;
- Access to genetic services “where people live;”
- Concerns about ethics;
- Medical providers’ limited knowledge about genetics.

**IDPH Supportive Efforts**

A key question for this needs assessment project was: *What could the Illinois Department of Public Health do to better support your efforts with respect to the provision of genetic services?* Responses generally fell into one of the following categories:

- **Funding** - Continue financial support (grants); offer more grants; provide more resources and more counseling services at the local level;
- **Health department staff education** – there were many compliments about the quality of the annual IDPH nursing genetics conference; however, respondents repeatedly stressed the need for more frequent (and shorter) education and training opportunities throughout the year, especially at the local level. As one respondent noted: “It is hard to travel to Chicago with the staff shortage and be gone for five days.” Respondents asked IDPH to consider downstate conferences, tele-conferences, videos, etc. Another suggestion was for IDPH to provide updates on already available educational resources and materials;
- **Public education** – a need for more pamphlets and other educational materials, including bilingual materials. One respondent stressed the importance and potential impact that community education would have on the health of the community.

**Conclusions**

Of the 71 health departments that had a staff member respond to the survey, 50 provided some level of genetic services; thirty-nine of those received grants from IDPH. Among the health departments that provided genetic services, there was variation between those that did and did not receive IDPH grant funding. A larger proportion of funded health departments used the IDPH genetic screening tool and provided on-site genetic clinics, referrals to genetic providers, and public and professional genetics education; a larger proportion also reported that they were able to meet their clients’ needs. When asked about the future impact of genetics on their public health programs, a greater proportion of funded than non-funded health departments foresaw a large impact, regardless of their provision of genetic services. Funded health departments were more comfortable with providing a variety of different services than were their non-funded counterparts, a finding that reveals areas where health department staff education can be targeted. The large proportion of respondents who did not know what public health programs areas would need to incorporate genetics in the next 3-5 years also indicates a need and opportunity for educational interventions. Respondents repeatedly raised the need for public and client education regarding genetic services. They indicated that IDPH should play a role in
funding genetic services (counseling), health department staff education, and community education.
Chapter 5: Survey Findings: Genetic Service Providers

The response rate for genetic service providers was 52% (78/150). The survey questionnaire is included in Appendix 3. Three respondents indicated that they were not currently practicing in genetics, so they were omitted from all analyses. Of the remaining 75 respondents, 60% (45) were genetic counselors, 35% (26) were MD and/or PhD geneticists, and 5% (4) fell into another category. Demographics, education, professional practice, and work patterns are described for all 75 respondents. The next set of analyses is limited to patient care genetic providers (n=60; 40 genetic counselors, 19 MD/PhD geneticists, and 1 other). The final section concludes with a brief summary of the responses of laboratory geneticists’ (n=19).

All Respondents

Demographic Characteristics

This section describes the gender, age, race and ethnicity of genetic providers who responded to the survey. Almost all genetic counselors and over half of MD/PhD geneticists were women (Table 5.1). Only one respondent was Hispanic, and 93% were white/Caucasian (Table 5.2); to preserve confidentiality, race and ethnicity are not presented separately for the two subgroups.

The median age of genetic counselors was 32 years, with a range of 26 to 65 years. Sixty percent of genetic counselors were under 35 years of age, and 96% were younger than 54 years. The age distribution of MD/PhD geneticists was older, with a median of 53 years. The youngest MD/PhD geneticist respondent was 41 years, and 54% were between 45-54 years. Thirty-four percent were at or approaching retirement age (55 and older) (Table 5.3).

Table 5.1 Gender of Genetic Service Provider Respondents

<table>
<thead>
<tr>
<th>Gender</th>
<th>Genetic Providers (n=75)</th>
<th>MD/PhD Geneticists (n=26)</th>
<th>Genetic Counselors (n=45)</th>
</tr>
</thead>
<tbody>
<tr>
<td>Women</td>
<td>83%</td>
<td>58%</td>
<td>96%</td>
</tr>
<tr>
<td>Men</td>
<td>17%</td>
<td>42%</td>
<td>4%</td>
</tr>
</tbody>
</table>

Table 5.2 Racial and Ethnic Representation of Genetic Service Provider Respondents

<table>
<thead>
<tr>
<th>Race/Ethnicity</th>
<th>Genetic Providers</th>
</tr>
</thead>
<tbody>
<tr>
<td>Asian</td>
<td>5%</td>
</tr>
<tr>
<td>Black/African American</td>
<td>1%</td>
</tr>
<tr>
<td>White</td>
<td>93%</td>
</tr>
<tr>
<td>Hispanic</td>
<td>1%</td>
</tr>
</tbody>
</table>
Table 5.3 Age Distribution of Genetic Service Provider Respondents

<table>
<thead>
<tr>
<th>Age Group</th>
<th>Genetic Providers (n=75)</th>
<th>MD/PhD Geneticists (n=26)</th>
<th>Genetic Counselors (n=45)</th>
</tr>
</thead>
<tbody>
<tr>
<td>Under 35 years</td>
<td>37%</td>
<td>0%</td>
<td>60%</td>
</tr>
<tr>
<td>35-44 years</td>
<td>23%</td>
<td>12%</td>
<td>29%</td>
</tr>
<tr>
<td>45-54 years</td>
<td>24%</td>
<td>54%</td>
<td>7%</td>
</tr>
<tr>
<td>55-64 years</td>
<td>8%</td>
<td>19%</td>
<td>0%</td>
</tr>
<tr>
<td>65 years or Older</td>
<td>8%</td>
<td>15%</td>
<td>4%</td>
</tr>
<tr>
<td>Median Age</td>
<td>39 yrs</td>
<td>53 yrs</td>
<td>32yrs</td>
</tr>
<tr>
<td>Mean Age</td>
<td>42 yrs</td>
<td>54 yrs</td>
<td>35 yrs</td>
</tr>
</tbody>
</table>

Nationally, most MD geneticists complete graduate medical education (GME) training in another specialty. Table 5.4 shows that the most common specialties reported were pediatrics (59%), other specialties (23%), and internal medicine and pathology (18% each).

Table 5.4 GME Training of MD Geneticists

<table>
<thead>
<tr>
<th>Specialty</th>
<th>Percent of MD Geneticists</th>
</tr>
</thead>
<tbody>
<tr>
<td>Pediatrics</td>
<td>59%</td>
</tr>
<tr>
<td>Internal Medicine</td>
<td>18%</td>
</tr>
<tr>
<td>Obstetrics/Gynecology</td>
<td>5%</td>
</tr>
<tr>
<td>Pathology</td>
<td>18%</td>
</tr>
<tr>
<td>Other Specialty</td>
<td>23%</td>
</tr>
<tr>
<td>No Other Specialty</td>
<td>0%</td>
</tr>
</tbody>
</table>

n=22; Individuals may have more than one type of training, thus totals > 100%.

Professional Practice and Work Patterns

This section presents findings relevant to practice and work patterns. Topics include primary work setting, length of tenure in current position, hours worked per week, and time spent in various professional activities.

The most common primary work setting for both MD/PhD geneticists and genetic counselors is the academic medical center (54% overall), followed by hospitals (26%) and medical practices (13%, single and multiple specialty grouped together) (Table 5.5). Few genetic service providers reported working in the other categories. Thirty-eight percent of MD/PhD geneticists had more than 25 years of tenure, with 96% having 6 or more (Table 5.6). Almost half of genetic counselors had been at their current work setting for five or fewer years.
Table 5.5 Primary Work Setting

<table>
<thead>
<tr>
<th>Setting</th>
<th>Genetic Providers (n=75)</th>
<th>MD/PhD Geneticists (n=26)</th>
<th>Genetic Counselors (n=45)</th>
</tr>
</thead>
<tbody>
<tr>
<td>Academic Medical Center/University</td>
<td>54%</td>
<td>52%</td>
<td>56%</td>
</tr>
<tr>
<td>Hospital</td>
<td>26%</td>
<td>28%</td>
<td>22%</td>
</tr>
<tr>
<td>Commercial Laboratory</td>
<td>3%</td>
<td>0%</td>
<td>4%</td>
</tr>
<tr>
<td>Medical Practice</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Managed Care Organization/HMO</td>
<td>0%</td>
<td>0%</td>
<td>0%</td>
</tr>
<tr>
<td>Medical Practice – Single Specialty</td>
<td>5%</td>
<td>8%</td>
<td>4%</td>
</tr>
<tr>
<td>Medical Practice – Multiple Specialty</td>
<td>8%</td>
<td>4%</td>
<td>11%</td>
</tr>
<tr>
<td>Government Agency (Non-Military)</td>
<td>0%</td>
<td>0%</td>
<td>0%</td>
</tr>
<tr>
<td>Pharmaceutical/Biotechnology</td>
<td>3%</td>
<td>4%</td>
<td>2%</td>
</tr>
<tr>
<td>Consulting (Group or Self-Employed)</td>
<td>0%</td>
<td>0%</td>
<td>0%</td>
</tr>
<tr>
<td>U.S. Military</td>
<td>0%</td>
<td>0%</td>
<td>0%</td>
</tr>
<tr>
<td>Other Setting</td>
<td>1%</td>
<td>4%</td>
<td>0%</td>
</tr>
</tbody>
</table>

Table 5.6 Tenure in Current Work Setting

<table>
<thead>
<tr>
<th>Tenure in Current Work Setting</th>
<th>Genetic Providers (n=75)</th>
<th>MD/PhD Geneticists (n=26)</th>
<th>Genetic Counselors (n=45)</th>
</tr>
</thead>
<tbody>
<tr>
<td>Less than 3 years</td>
<td>9%</td>
<td>0%</td>
<td>16%</td>
</tr>
<tr>
<td>3-5 years</td>
<td>20%</td>
<td>4%</td>
<td>31%</td>
</tr>
<tr>
<td>6-10 years</td>
<td>24%</td>
<td>27%</td>
<td>24%</td>
</tr>
<tr>
<td>11-15 years</td>
<td>15%</td>
<td>8%</td>
<td>18%</td>
</tr>
<tr>
<td>16-20 years</td>
<td>12%</td>
<td>15%</td>
<td>7%</td>
</tr>
<tr>
<td>21-25 years</td>
<td>5%</td>
<td>8%</td>
<td>4%</td>
</tr>
<tr>
<td>More than 25 years</td>
<td>15%</td>
<td>38%</td>
<td>0%</td>
</tr>
</tbody>
</table>

Respondents were asked to indicate the percentage of their time spent in a typical week in a variety of professional activities (Table 5.7). Note that these numbers reflect aggregate time distribution across all respondents. MD/PhD geneticists spent 39% of their time in patient care, and about equal amounts of time in laboratory, research, and administration activities. Genetic counselors spend substantially more time in patient care activities (61%). Teaching was the most common reported activity (64 genetic providers), followed by clinical patient care (60), administration (51), and clinical or other research (37).
Table 5.7 Aggregate Percent of Time Spent in Professional Activities

<table>
<thead>
<tr>
<th>Professional Activity</th>
<th>Total</th>
<th>MD/PhD Geneticists</th>
<th>Genetic Counselors</th>
</tr>
</thead>
<tbody>
<tr>
<td>Clinical Patient Care</td>
<td>51%</td>
<td>39%</td>
<td>61%</td>
</tr>
<tr>
<td>Clinical Laboratory</td>
<td>10%</td>
<td>15%</td>
<td>5%</td>
</tr>
<tr>
<td>Research</td>
<td>10%</td>
<td>14%</td>
<td>8%</td>
</tr>
<tr>
<td>Administration</td>
<td>12%</td>
<td>13%</td>
<td>10%</td>
</tr>
<tr>
<td>Teaching</td>
<td>9%</td>
<td>10%</td>
<td>9%</td>
</tr>
<tr>
<td>Writing</td>
<td>2%</td>
<td>5%</td>
<td>1%</td>
</tr>
<tr>
<td>Other</td>
<td>5%</td>
<td>4%</td>
<td>6%</td>
</tr>
</tbody>
</table>

Respondents were asked to report the average number of hours they worked in a typical week (Table 5.8). MD/PhD geneticists worked almost 51 more hours per week (median=58 hours) and genetic counselors worked about 40 hours per week (median=40 hours). Forty-eight percent of MD/PhD geneticists reported working 60 or more hours per week, with another 20% working 50-59 hours per week.

Table 5.8 Hours Worked in Typical Week

<table>
<thead>
<tr>
<th>Hours Worked/Week</th>
<th>Genetic Providers (n=74)</th>
<th>MD/PhD Geneticists (n=25)</th>
<th>Genetic Counselors (n=45)</th>
</tr>
</thead>
<tbody>
<tr>
<td>1-19 hrs</td>
<td>7%</td>
<td>8%</td>
<td>7%</td>
</tr>
<tr>
<td>20-29 hrs</td>
<td>8%</td>
<td>4%</td>
<td>9%</td>
</tr>
<tr>
<td>30-39 hrs</td>
<td>9%</td>
<td>8%</td>
<td>11%</td>
</tr>
<tr>
<td>40 hrs</td>
<td>26%</td>
<td>12%</td>
<td>33%</td>
</tr>
<tr>
<td>41-49 hrs</td>
<td>12%</td>
<td>0%</td>
<td>20%</td>
</tr>
<tr>
<td>50-59 hrs</td>
<td>18%</td>
<td>20%</td>
<td>16%</td>
</tr>
<tr>
<td>60 hrs or more</td>
<td>20%</td>
<td>48%</td>
<td>4%</td>
</tr>
<tr>
<td>Median</td>
<td>41 hrs</td>
<td>58 hrs</td>
<td>40 hrs</td>
</tr>
<tr>
<td>Mean</td>
<td>43.6 hrs</td>
<td>50.7 hrs</td>
<td>39.8 hrs</td>
</tr>
</tbody>
</table>

We asked respondents to estimate the number of lectures or presentations they gave to the following types of audiences over the previous 12 months. Of the 74 respondents, only 5 indicated that they did not spend any time teaching. By far, the most common audience was medical students/residents/fellows.
Table 5.9 Total Number of Lectures/Presentations by Type of Audience

<table>
<thead>
<tr>
<th>Audience</th>
<th>Genetic Providers</th>
<th>MD/PhD Geneticists</th>
<th>Genetic Counselors</th>
</tr>
</thead>
<tbody>
<tr>
<td>Educators (teachers, professors)</td>
<td>78</td>
<td>56</td>
<td>20</td>
</tr>
<tr>
<td>General public</td>
<td>64</td>
<td>33</td>
<td>29</td>
</tr>
<tr>
<td>Medical students, residents, fellows</td>
<td>672</td>
<td>299</td>
<td>362</td>
</tr>
<tr>
<td>Other health professionals</td>
<td>166</td>
<td>89</td>
<td>66</td>
</tr>
<tr>
<td>Support groups, disease organizations</td>
<td>31</td>
<td>16</td>
<td>14</td>
</tr>
<tr>
<td>Students (college, graduate)</td>
<td>133</td>
<td>21</td>
<td>108</td>
</tr>
<tr>
<td>Other</td>
<td>3</td>
<td>0</td>
<td>3</td>
</tr>
</tbody>
</table>

Patient Care Genetic Providers

This section focuses on the 60 respondents who reported providing face-to-face patient care. As above, results are shown for the subgroups of MD/PhD geneticists and genetic counselors.

The number of hours worked each week was similar to that of all respondents. When excluding non-patient care providers, the percentage of time spent on clinical patient care increased and time spent in the clinical laboratory decreased (Table 5.10).

Table 5.10 Aggregate Percent of Time Spent in Professional Activities, Patient Care Providers

<table>
<thead>
<tr>
<th>Professional Activity</th>
<th>Total</th>
<th>MD/PhD Geneticists</th>
<th>Genetic Counselors</th>
</tr>
</thead>
<tbody>
<tr>
<td>Clinical Patient Care</td>
<td>64%</td>
<td>54%</td>
<td>68%</td>
</tr>
<tr>
<td>Clinical Laboratory</td>
<td>3%</td>
<td>7%</td>
<td>1%</td>
</tr>
<tr>
<td>Research</td>
<td>7%</td>
<td>11%</td>
<td>6%</td>
</tr>
<tr>
<td>Administration</td>
<td>10%</td>
<td>12%</td>
<td>10%</td>
</tr>
<tr>
<td>Teaching</td>
<td>10%</td>
<td>11%</td>
<td>10%</td>
</tr>
<tr>
<td>Writing</td>
<td>2%</td>
<td>6%</td>
<td>1%</td>
</tr>
<tr>
<td>Other</td>
<td>3%</td>
<td>0%</td>
<td>4%</td>
</tr>
</tbody>
</table>

Respondents were asked about the staffing levels at their primary work site as well as the adequacy of staffing. Due to a large amount of missing and incomplete responses, we are unable to report on FTE staffing levels. However, half of respondents felt there were too few MD geneticists and genetic counselors, 43% cited too few laboratory assistants/support staff, and 31% said too few “other” professionals (Table 5.11). Having too much staff on site was not noted as a problem.
Table 5.11 Adequacy of Staffing Levels at Primary Genetics Clinical Site

<table>
<thead>
<tr>
<th>Staff</th>
<th>Too Few</th>
<th>Right Number</th>
<th>Too Many</th>
<th>Don’t Know</th>
</tr>
</thead>
<tbody>
<tr>
<td>MD Geneticists</td>
<td>51%</td>
<td>46%</td>
<td>0%</td>
<td>4%</td>
</tr>
<tr>
<td>PhD Geneticists</td>
<td>10%</td>
<td>63%</td>
<td>0%</td>
<td>27%</td>
</tr>
<tr>
<td>Genetic Counselors</td>
<td>51%</td>
<td>49%</td>
<td>0%</td>
<td>0%</td>
</tr>
<tr>
<td>Lab. Assts./ Support Staff</td>
<td>43%</td>
<td>46%</td>
<td>0%</td>
<td>11%</td>
</tr>
<tr>
<td>Technologists/technicians</td>
<td>10%</td>
<td>64%</td>
<td>0%</td>
<td>26%</td>
</tr>
<tr>
<td>Other professionals</td>
<td>31%</td>
<td>38%</td>
<td>3%</td>
<td>28%</td>
</tr>
</tbody>
</table>

Genetic providers were asked to estimate the number of new outpatient, follow-up outpatient, and inpatient patient visits they had in the past 12 months. They were also asked to estimate the typical wait time for a new and follow-up patient, the number and percentage of missed appointments each week, frequency with which issues were discussed with patients, and whether their practice was full.

There was a wide range of reported annual patient visits, both by visit type and provider type (Table 5.12). MD/PhD geneticists had a higher average number of patient visits for all visit types than genetic counselors, although the median number of new outpatient visits was higher for genetic counselors. Patient care genetic providers reported an average of 12% of missed appointments each week, with MD/PhD geneticists reporting about double this amount compared to genetic counselors (data not shown). Most new patients had to wait one-to-four weeks for an appointment, somewhat less to see a genetic counselor versus a geneticist (Table 5.13). The wait for existing patients was somewhat shorter than for new patients (Table 5.14).

Table 5.12 Estimated Annual Patient Visits

<table>
<thead>
<tr>
<th>Annual Patient Visits</th>
<th>Patient Care Genetic Providers (n=59)</th>
<th>MD/PhD Geneticists (n=18)</th>
<th>Genetic Counselors (n=40)</th>
</tr>
</thead>
<tbody>
<tr>
<td>New outpatient</td>
<td>Mean = 301.1&lt;br&gt;Median = 200&lt;br&gt;Range = 5-3000</td>
<td>Mean = 398.1&lt;br&gt;Median = 175&lt;br&gt;Range = 20-3000</td>
<td>Mean = 261.4&lt;br&gt;Median = 208&lt;br&gt;Range = 5-800</td>
</tr>
<tr>
<td>Follow-up Outpatient</td>
<td>Mean = 105.9&lt;br&gt;Median = 70&lt;br&gt;Range = 0-500</td>
<td>Mean = 131.4&lt;br&gt;Median = 100&lt;br&gt;Range = 30-450</td>
<td>Mean = 87.8&lt;br&gt;Median = 35&lt;br&gt;Range = 0-500</td>
</tr>
<tr>
<td>Inpatient</td>
<td>Mean = 30.9&lt;br&gt;Median = 5.5&lt;br&gt;Range = 0-240</td>
<td>Mean = 66.5&lt;br&gt;Median = 50&lt;br&gt;Range = 0-240</td>
<td>Mean = 13.9&lt;br&gt;Median = 5&lt;br&gt;Range = 0-100</td>
</tr>
</tbody>
</table>
Table 5.13 Typical Waiting Time for a New Patient Appointment

<table>
<thead>
<tr>
<th>Typical Waiting Time for an Appointment for a New Patient</th>
<th>Patient Care Genetic Providers (n=60)</th>
<th>MD/PhD Geneticists (n=19)</th>
<th>Genetic Counselors (n=40)</th>
</tr>
</thead>
<tbody>
<tr>
<td>0 (Same Day)</td>
<td>5%</td>
<td>0%</td>
<td>9%</td>
</tr>
<tr>
<td>1-2 Days</td>
<td>4%</td>
<td>5%</td>
<td>3%</td>
</tr>
<tr>
<td>3-6 Days</td>
<td>22%</td>
<td>5%</td>
<td>31%</td>
</tr>
<tr>
<td>1 Week – Less Than 1 Month</td>
<td>46%</td>
<td>53%</td>
<td>40%</td>
</tr>
<tr>
<td>1-2 Months</td>
<td>24%</td>
<td>37%</td>
<td>17%</td>
</tr>
</tbody>
</table>

Table 5.14 Typical Waiting Time for an Existing Patient Appointment

<table>
<thead>
<tr>
<th>Typical Waiting Time for an Appointment for an Existing Patient</th>
<th>Patient Care Genetic Providers (n=60)</th>
<th>MD/PhD Geneticists (n=19)</th>
<th>Genetic Counselors (n=40)</th>
</tr>
</thead>
<tbody>
<tr>
<td>0 (Same Day)</td>
<td>6%</td>
<td>0%</td>
<td>9%</td>
</tr>
<tr>
<td>1-2 Days</td>
<td>10%</td>
<td>17%</td>
<td>6%</td>
</tr>
<tr>
<td>3-6 Days</td>
<td>23%</td>
<td>6%</td>
<td>33%</td>
</tr>
<tr>
<td>1 Week – Less Than 1 Month</td>
<td>40%</td>
<td>44%</td>
<td>39%</td>
</tr>
<tr>
<td>1-2 Months</td>
<td>21%</td>
<td>33%</td>
<td>12%</td>
</tr>
</tbody>
</table>

Very few genetic providers reported that they could not accept any new patients (Table 5.15). About one-fourth of genetic counselors and almost one-half of MD/PhD geneticists reported that their practice was far from full.

Table 5.15 Capacity to See New Patients

<table>
<thead>
<tr>
<th>Capacity to See New Patients</th>
<th>Patient Care Genetic Providers (n=60)</th>
<th>MD/PhD Geneticists (n=19)</th>
<th>Genetic Counselors (n=40)</th>
</tr>
</thead>
<tbody>
<tr>
<td>Cannot accept any more genetics patients, practice is full</td>
<td>8%</td>
<td>6%</td>
<td>9%</td>
</tr>
<tr>
<td>Can accept some new genetics patients, practice nearly full</td>
<td>62%</td>
<td>50%</td>
<td>68%</td>
</tr>
<tr>
<td>Can accept many new genetics patients practice far from full</td>
<td>31%</td>
<td>47%</td>
<td>24%</td>
</tr>
</tbody>
</table>

Almost all genetic providers discussed confidentiality of test results, potential of inconclusive results, testing fees and insurance coverage, and the technical accuracy of the test with all of their patients (Table 5.16). The risks of employment and insurance discrimination were less frequently discussed. For almost all of these issues, more MD/PhD geneticists discussed them with all of their patients than did genetic counselors.
Table 5.16 Percent of Geneticists and Genetic Counselors that Discuss Specific Risks Associated with Genetic Testing

<table>
<thead>
<tr>
<th>Issues Discussed with Patients</th>
<th>None</th>
<th>Few</th>
<th>Some</th>
<th>Many</th>
<th>All</th>
</tr>
</thead>
<tbody>
<tr>
<td>Confidentiality of Test Results</td>
<td>0%</td>
<td>0%</td>
<td>6%</td>
<td>3%</td>
<td>91%</td>
</tr>
<tr>
<td>Risks of Employment Discrimination</td>
<td>6%</td>
<td>6%</td>
<td>3%</td>
<td>33%</td>
<td>52%</td>
</tr>
<tr>
<td>Risks of Insurance Discrimination</td>
<td>3%</td>
<td>0%</td>
<td>3%</td>
<td>27%</td>
<td>67%</td>
</tr>
<tr>
<td>Potential of Inconclusive Results</td>
<td>0%</td>
<td>0%</td>
<td>3%</td>
<td>12%</td>
<td>85%</td>
</tr>
<tr>
<td>Psychological Implications of Test Results</td>
<td>3%</td>
<td>0%</td>
<td>6%</td>
<td>13%</td>
<td>78%</td>
</tr>
<tr>
<td>Testing Fees and Insurance Coverage</td>
<td>3%</td>
<td>0%</td>
<td>3%</td>
<td>6%</td>
<td>88%</td>
</tr>
<tr>
<td>Technical Accuracy of Test</td>
<td>3%</td>
<td>0%</td>
<td>3%</td>
<td>9%</td>
<td>85%</td>
</tr>
<tr>
<td>Right to Choose Not to Receive Test Results</td>
<td>3%</td>
<td>3%</td>
<td>3%</td>
<td>10%</td>
<td>80%</td>
</tr>
</tbody>
</table>

N=35 (Those that responded that they see patients undergoing genetic susceptibility/presymptomatic testing)

Most referrals to genetic providers were from specialist physicians, followed by generalist physicians. A greater percentage of referrals to genetic counselors were from specialist physicians than were referrals to MD/PhD geneticists (Table 5.17).

Table 5.17 Percentage of Referrals Received from Listed Sources

<table>
<thead>
<tr>
<th>Referral Source</th>
<th>Patient Care Genetic Providers (n=60)</th>
<th>MD/PhD Geneticists (n=19)</th>
<th>Genetic Counselors (n=40)</th>
</tr>
</thead>
<tbody>
<tr>
<td>Generalist physicians</td>
<td>24%</td>
<td>34%</td>
<td>21%</td>
</tr>
<tr>
<td>Specialist physicians</td>
<td>49%</td>
<td>40%</td>
<td>57%</td>
</tr>
<tr>
<td>MD/DO geneticists</td>
<td>6%</td>
<td>3%</td>
<td>5%</td>
</tr>
<tr>
<td>Patient Self-Referral</td>
<td>8%</td>
<td>10%</td>
<td>7%</td>
</tr>
<tr>
<td>Genetic Counselors</td>
<td>3%</td>
<td>3%</td>
<td>3%</td>
</tr>
<tr>
<td>IDPH Genetics and Newborn Screening</td>
<td>4%</td>
<td>3%</td>
<td>2%</td>
</tr>
<tr>
<td>Genetic testing laboratories or programs</td>
<td>3%</td>
<td>2%</td>
<td>1%</td>
</tr>
<tr>
<td>Local health departments</td>
<td>3%</td>
<td>4%</td>
<td>3%</td>
</tr>
<tr>
<td>Other</td>
<td>0%</td>
<td>0%</td>
<td>0%</td>
</tr>
</tbody>
</table>

Twenty-one respondents (8 MD/PhD geneticists and 13 genetic counselors) indicated that they provided between 2 and 52 outreach sessions per year (median = 12). Genetic counselors averaged 18 annual outreach sessions, while MD/PhD geneticists averaged 14. The identified settings for providing outreach services were hospitals (8), health departments (7), academic medical centers (4), multi-specialty medical practices and community health centers (both 3), and single-specialty medical practices (2).
Patient Sociodemographic Characteristics and Insurance Status

Race and Ethnicity Of Patients

We separated race (African American, White/Caucasian, etc.) and ethnicity (Hispanic/not Hispanic) into two questions but many respondents categorized Hispanic as “other” under the question concerning race. Therefore, the responses were not consistent and we cannot report accurate findings. However, we can estimate that around 60% of patients were White/Caucasian and about 17% were African American; approximately one-fourth of patients were Hispanic.

Patients by Age/Type

Genetic counselors saw a larger proportion of prenatal/reproductive patients (42%), and fewer infants, children, and adolescents (22%), than did MD/PhD geneticists (19% and 55%) (Table 5.18). Genetic counselors also had a larger caseload of carrier screening and adult patients with non-reproductive concerns.

Table 5.19 breaks down patient caseload by category of genetic referral. As shown in Tables 5.18 and 5.19, a large proportion of genetic counselors’ caseload consists of patients with prenatal/reproductive issues. About 34% of patients seen by geneticists are seen for dysmorphology/syndromes/birth defects (Table 5.19), followed by cancer (18%), prenatal/reproductive issues (17%), and metabolic conditions (13%).

Table 5.18 Patients by Age/Type

<table>
<thead>
<tr>
<th>Patient Type</th>
<th>Patient Care Genetic Providers (n=60)</th>
<th>MD/PhD Geneticists (n=19)</th>
<th>Genetic Counselors (n=40)</th>
</tr>
</thead>
<tbody>
<tr>
<td>Newborns and infants up to one year of age</td>
<td>15%</td>
<td>26%</td>
<td>10%</td>
</tr>
<tr>
<td>Children and adolescents</td>
<td>18%</td>
<td>29%</td>
<td>12%</td>
</tr>
<tr>
<td>Prenatal/Reproductive</td>
<td>35%</td>
<td>19%</td>
<td>42%</td>
</tr>
<tr>
<td>Carrier Screening</td>
<td>7%</td>
<td>4%</td>
<td>8%</td>
</tr>
<tr>
<td>Adults (non reproductive)</td>
<td>25%</td>
<td>22%</td>
<td>27%</td>
</tr>
</tbody>
</table>
Table 5.19 Patients by Genetic Referral Category

<table>
<thead>
<tr>
<th>Patient Groups</th>
<th>Patient Care Genetic Providers (n=60)</th>
<th>MD/PhD Geneticists (n=19)</th>
<th>Genetic Counselors (n=40)</th>
</tr>
</thead>
<tbody>
<tr>
<td>Dysmorphology/syndromes/birth defects</td>
<td>17%</td>
<td>34%</td>
<td>11%</td>
</tr>
<tr>
<td>Developmental delay/mental retardation</td>
<td>2%</td>
<td>4%</td>
<td>2%</td>
</tr>
<tr>
<td>Metabolic conditions</td>
<td>11%</td>
<td>13%</td>
<td>8%</td>
</tr>
<tr>
<td>Prenatal/Reproductive issues</td>
<td>40%</td>
<td>17%</td>
<td>49%</td>
</tr>
<tr>
<td>Cancer</td>
<td>16%</td>
<td>18%</td>
<td>15%</td>
</tr>
<tr>
<td>Common/complex disorders</td>
<td>3%</td>
<td>3%</td>
<td>3%</td>
</tr>
<tr>
<td>Adult-onset single gene disorders</td>
<td>6%</td>
<td>4%</td>
<td>7%</td>
</tr>
<tr>
<td>Other</td>
<td>5%</td>
<td>7%</td>
<td>5%</td>
</tr>
</tbody>
</table>

Patient care genetic providers reported that 35% of their patients were covered under managed care contracts, 20% were privately insured, and 27% had Medicaid coverage; only 5% were uninsured (Table 5.20). Genetic counselors saw a much greater proportion of managed care patients than MD/PhD geneticists, who saw more uninsured patients.

Table 5.20 Insurance Coverage of Patients

<table>
<thead>
<tr>
<th>Patient Insurance Coverage</th>
<th>Patient Care Genetic Providers (n=60)</th>
<th>MD/PhD Geneticists (n=19)</th>
<th>Genetic Counselors (n=40)</th>
</tr>
</thead>
<tbody>
<tr>
<td>Managed Care</td>
<td>35%</td>
<td>25%</td>
<td>40%</td>
</tr>
<tr>
<td>Private Insurance</td>
<td>20%</td>
<td>23%</td>
<td>19%</td>
</tr>
<tr>
<td>Medicaid</td>
<td>27%</td>
<td>26%</td>
<td>27%</td>
</tr>
<tr>
<td>Medicare</td>
<td>5%</td>
<td>7%</td>
<td>4%</td>
</tr>
<tr>
<td>Military</td>
<td>1%</td>
<td>2%</td>
<td>1%</td>
</tr>
<tr>
<td>Uninsured</td>
<td>5%</td>
<td>10%</td>
<td>3%</td>
</tr>
<tr>
<td>Unknown</td>
<td>6%</td>
<td>8%</td>
<td>6%</td>
</tr>
</tbody>
</table>

Telemedicine

We asked respondents about their views and use of telemedicine techniques to provide genetic diagnosis and/or counseling. Few genetic providers had used telemedicine, but those who had used it all felt it was somewhat effective. MD/PhD geneticists were more supportive of its potential than were genetic counselors (Table 5.21).
Table 5.21 Views and Use of Telemedicine Techniques to Provide Genetic Diagnosis and/or Counseling

<table>
<thead>
<tr>
<th>Patient Care Genetic Providers (n=60)</th>
<th>MD/PhD Geneticists (n=19)</th>
<th>Genetic Counselors (n=40)</th>
</tr>
</thead>
<tbody>
<tr>
<td>Yes, I believe this is an effective method of service delivery</td>
<td>0%</td>
<td>0%</td>
</tr>
<tr>
<td>Yes, I think this method of service delivery is somewhat effective</td>
<td>10%</td>
<td>16%</td>
</tr>
<tr>
<td>Yes, but I do not think this is an effective method of service delivery</td>
<td>0%</td>
<td>0%</td>
</tr>
<tr>
<td>No, I have not used but think this might be an effective method of service delivery</td>
<td>43%*</td>
<td>53%</td>
</tr>
<tr>
<td>No, I have not used but think this would not be an effective way to deliver genetic services</td>
<td>17%</td>
<td>26%</td>
</tr>
<tr>
<td>No, but I do provide diagnostic and/or counseling services by telephone</td>
<td>29%</td>
<td>0%</td>
</tr>
</tbody>
</table>

*One respondent noted both: “No, I have not used but think this might be an effective method of service delivery,” and “No, but I do provide diagnostic and/or counseling services by telephone.”

Laboratory Geneticists & Clinical Genetics Laboratory Services

There were 19 respondents who indicated that they worked in a clinical genetics laboratory or provide clinical genetic laboratory services; of those, 11 were MD/PhD geneticists, 6 were genetic counselors, and 2 were other professionals. Twelve (8 MD/PhD geneticists and 4 genetic counselors) of the 19 also provided face-to-face patient care. Seventeen of these providers worked in one laboratory each, the other two worked in two laboratories.

This group can be summarized as follows:

- 74% were women; including all of the genetic counselors and other professionals, as were 55% of the geneticists;
- 95% (18/19) were White/Caucasian, and 100% were non-Hispanic;
- Their age distribution was similar to that of all genetic provider respondents;
- Of the MD geneticists, 8 had completed GME training in pediatrics and 1 in pathology;
- 28% reported that their primary work setting was in an academic medical center/university, 28% in a hospital, 34% in a single- or multiple-specialty medical practice, and the remaining 11% in a commercial laboratory;
- 72% of geneticists had been at their current work setting for 16 or more years; 67% of genetic counselors had been at their current work setting for fewer than 5 years;
- 49% worked 50 or more hours per week; the mean was 52.6 hours for geneticists and 38.8 hours for genetic counselors;
- Even for this group, 40% of their time was spent in clinical patient care, followed by 33% in clinical laboratory activities, 13% in administration, and 7% teaching (see Table 22). By number of respondents, 16 laboratory geneticists reported spending at least some time in
clinical laboratory care, 15 in administration, 13 in teaching, 12 in clinical patient care, 6 in clinical or other research, and 5 in writing;

- The most common area that providers spent time working in was clinical cytogenetics (74% of clinical laboratory time) followed by clinical molecular genetics (10%), prenatal screening (11%), and clinical biochemical genetics (3%; Table 22);
- The most common clinical laboratory activities included communicating with non-genetics healthcare professionals (21% of time), results interpretation/reports (39%), communicating with genetics professionals (7%), case research and management (10%), communicating with patients (13%), and test-related activities (10%; Table 5.22);

### Table 5.22 Laboratory Geneticist Time Spent in Professional Activities

<table>
<thead>
<tr>
<th>Time spent in:</th>
<th>% of time</th>
<th>Number of Respondents Reporting Any Time</th>
</tr>
</thead>
<tbody>
<tr>
<td>Clinical Patient Care</td>
<td>40%</td>
<td>12</td>
</tr>
<tr>
<td>Clinical Laboratory</td>
<td>33%</td>
<td>16</td>
</tr>
<tr>
<td>Research</td>
<td>5%</td>
<td>6</td>
</tr>
<tr>
<td>Administration</td>
<td>13%</td>
<td>15</td>
</tr>
<tr>
<td>Teaching</td>
<td>7%</td>
<td>13</td>
</tr>
<tr>
<td>Writing</td>
<td>2%</td>
<td>5</td>
</tr>
<tr>
<td>Clinical laboratory time spent in:</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Clinical Biochemical Genetics</td>
<td>3%</td>
<td>5</td>
</tr>
<tr>
<td>Clinical Molecular Genetics</td>
<td>10%</td>
<td>10</td>
</tr>
<tr>
<td>Clinical Cytogenetics</td>
<td>74%</td>
<td>16</td>
</tr>
<tr>
<td>Prenatal Screening (AFP, Multiple Marker)</td>
<td>11%</td>
<td>8</td>
</tr>
<tr>
<td>Newborn Screening</td>
<td>0%</td>
<td>1</td>
</tr>
<tr>
<td>Non-Genetics Laboratories</td>
<td>2%</td>
<td>1</td>
</tr>
<tr>
<td>Of clinical laboratory activities, time spent in:</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Test-related activities (benchwork, assay development, assay QC)</td>
<td>10%</td>
<td>9</td>
</tr>
<tr>
<td>Results interpretation/reports (review/approve assays, reports, etc.)</td>
<td>39%</td>
<td>15</td>
</tr>
<tr>
<td>Case research/management, follow-up</td>
<td>10%</td>
<td>14</td>
</tr>
<tr>
<td>Communication with genetic professionals</td>
<td>7%</td>
<td>15</td>
</tr>
<tr>
<td>Communication with other (non-genetics) healthcare professionals</td>
<td>21%</td>
<td>18</td>
</tr>
<tr>
<td>Communication with patients</td>
<td>13%</td>
<td>12</td>
</tr>
</tbody>
</table>

- When asked about the types of test analyzed by patient type, 15 providers each reported tests for prenatal/reproductive patients (52% of tests) and non-pregnant adults (17%), followed by newborns and infants (10%), children and adolescents (13%), and carrier screening (7%; Table 5.23);
### Table 5.23 Tests by Patient Type

<table>
<thead>
<tr>
<th>Types of tests analyzed by patient type:</th>
<th>% of Tests</th>
<th>Number of Respondents Reporting Any Tests</th>
</tr>
</thead>
<tbody>
<tr>
<td>Newborns and Infants (less than one year of age)</td>
<td>10%</td>
<td>13</td>
</tr>
<tr>
<td>Children and Adolescents (not pregnant)</td>
<td>13%</td>
<td>12</td>
</tr>
<tr>
<td>Prenatal/Reproductive genetics patients</td>
<td>52%</td>
<td>15</td>
</tr>
<tr>
<td>Carrier Screening</td>
<td>7%</td>
<td>9</td>
</tr>
<tr>
<td>Adults (not pregnant)</td>
<td>17%</td>
<td>15</td>
</tr>
</tbody>
</table>

- Most laboratory providers reported receiving samples submitted by specialist physicians (51% of samples submitted), generalist physicians (17%), medical geneticists (14%), and genetic counselors (14%; Table 5.24).

### Table 5.24 Samples Submitted by Type of Provider

<table>
<thead>
<tr>
<th>Samples submitted by:</th>
<th>% of Samples</th>
<th>Number of Respondents Reporting Any Samples</th>
</tr>
</thead>
<tbody>
<tr>
<td>MD/DO geneticists</td>
<td>14%</td>
<td>12</td>
</tr>
<tr>
<td>Generalist physicians (pediatricians, internists, family practitioners)</td>
<td>17%</td>
<td>13</td>
</tr>
<tr>
<td>Specialist physicians (OBGYN, oncology, neurology)</td>
<td>51%</td>
<td>15</td>
</tr>
<tr>
<td>Genetic counselors</td>
<td>14%</td>
<td>11</td>
</tr>
<tr>
<td>Patient (self-referral or family referral)</td>
<td>2%</td>
<td>6</td>
</tr>
<tr>
<td>Other genetic testing laboratories or programs</td>
<td>2%</td>
<td>4</td>
</tr>
<tr>
<td>IDPH Genetics and Newborn Screening</td>
<td>0%</td>
<td>1</td>
</tr>
<tr>
<td>Local health departments</td>
<td>1%</td>
<td>2</td>
</tr>
<tr>
<td>Other</td>
<td>0%</td>
<td>0</td>
</tr>
</tbody>
</table>

- Staffing data were incomplete, but most respondents indicated that staffing levels at their primary clinical genetics laboratory included the right number for MD geneticists, PhD geneticists, and genetic counselors.

### All Respondents

All respondents were asked about their familiarity with the IDPH Genetics and Newborn Screening Program, and were given an opportunity to discuss access barriers, unmet patient needs, major issues facing genetic service provision, and how IDPH could better support their efforts.

Overall, 37% of respondents reported being very familiar with Illinois’ Genetic and Newborn Screening Program, while 25% were familiar, 32% were somewhat familiar, and 5% were not familiar (Figure 5.1). For all respondents, patient care genetic providers, and clinical laboratory genetic providers, MD/PhD geneticists were more familiar with Illinois’ Genetic and Newborn Screening Program than were genetic counselors.
Barriers and Areas of Unmet Need

We asked respondents to identify barriers to patient access and/or areas of unmet need with respect to medical genetic services in Illinois. The responses can be grouped around the following themes:

- **Financial/Insurance Constraints** - lack of insurance coverage for genetic testing, counseling, or other services (and was often a factor in cancelled or missed appointments); cost of genetic tests; inadequate or non-existent insurance reimbursement; and lack of coverage for patients using Medicaid;

- **Lack of Physician Knowledge/Education/Awareness Regarding Genetic Services** – primary care physicians do not appear to have enough information about when to refer patients and to whom they should refer to (too few and/or inappropriate referrals); inadequate awareness of available resources; too little training about interpreting test results and ordering tests when appropriate; may not understand the benefit of referral to genetic services for the patient; lack of follow-up on family history; problems exacerbated in rural settings;

- **Workforce Size and Distribution** – too few medical geneticists; lack of genetic service providers outside of Chicago, large cities; lack of support for genetic counselors;
• Need for Public Education – lack of understanding of genetic issues, it’s value, and the availability of genetic services by the public; preconceived notions of what genetic counseling is about;
• Access issues – distance to where providers and services are located; lack of transportation; language barriers; cultural barriers; same access issues as for health care in general;
• Other barriers – fear of insurance discrimination; state government support; bureaucracy; and
• Other unmet needs – genetic evaluation for hearing loss, mental retardation, and congenital heart defects; follow-up of partners of pregnant women; public support for graduate education; adequate prenatal services.

Key Issues Facing Genetic Service Providers

We asked genetic service providers: In your opinion, what are the biggest issues genetic service providers in your field will face over the next 5 to 10 years with respect to the provision of genetic services in Illinois? Reimbursement, insurance, and financial concerns dominated the responses.

• Reimbursement, insurance, and financial concerns. Lack of reimbursement by insurance companies for genetic services and testing was a primary concern. A need for billing codes was noted, including reimbursement for the time to arrange and get pre-approval for genetic tests, for counseling services, and for obtaining informed consent. Another issue raised was that genetic counselors are currently not able to bill for their services. Some respondents noted concerns about the ability of their genetics program or department to stay “in business.” For example,
  ○ “I think reimbursement/billing issues are going to dramatically affect all genetics providers as those departments seem to struggle to get adequate monies to stay operational.”
  ○ “…concern that we will need to bring in enough money to cover the expenses of [our] genetics program.”
One provider expressed concern about cost and insurance, including a declining number of patients with insurance coverage, this way: “I fear ‘genetics for the wealthy.’”

• Geneticist workforce. There were concerns that there are currently, and will be in the future, an inadequate number of trained medical geneticists, which were related to other issues: the number of newly trained geneticists versus the number of retiring geneticists; genetic counselor access to medical geneticists; and whether the field can keep up with the increasing number of genetic tests. One respondent described an “inability to be financially secure under current reimbursement policies;” this sentiment may be related to the number of practicing geneticists. Respondents also expressed that continuing education is difficult given the rapid changes in the field, and that increased specialization may mean that some patients will be unable to obtain appointments with genetic service providers.

• Genetic counselor workforce. These issues included an inadequate number of genetic counselors, and the dependence of current genetic counselor positions on grant funding (state and other). The recent genetic counselor licensure act, described in Chapter 8, was mentioned regarding billing and reimbursement of counseling services, and potential impact on non-licensed genetics professionals, especially nurses.

• Changes in how genetics will be delivered and the role of genetic versus non-genetic service providers. Respondents discussed this theme in the following ways:
“Who will provide genetic services in the near future – genetic professionals or primary care physicians?”

“Maintaining their [genetic providers’] uniqueness in patient service providing – i.e., services will be assumed by pediatricians, obstetricians, ultrasound, and maternal fetal medicine.”

“Clarifying the lines of responsibility of genetic versus non-genetic professionals – what is our role and how should we divide up responsibilities.”

“How primary care physicians will be integrated into providing genetic services. This is beyond just education but the implementation.”

“Primary care physicians will provide genetic services with lack of knowledge in the field.”

“Genetic services will increasingly be specialty specific, i.e., tests related to cardiologist will be ordered by cardiologists, not genetic providers, unless they become part of US Preventive Task Force Recommendations164.”

“Genetic testing will become more ‘routine’ and move into the general physician’s arena. They are not fully educated enough to explain the risks, limitation, [and] benefits of the tests and aren’t always educated enough to know their limitations and refer their patients to a specialist.”

- **Education of non-genetic health professionals.** Many of the above statements demonstrate concerns genetic providers have about the level of education of other professionals – both primary care and specialty providers – and their role in the genetic service delivery system. Genetic providers expressed concerns about the increasing number of tests available and the difficulty in interpreting the test results, whether PCPs are properly counseling patients, and the appropriate use of genetic services and testing in general.

- **Access issues.** Concerns included: equal access for low socioeconomic groups; reaching underserved areas and populations; declining number of patients with insurance; demand exceeding provider supply; limited availability of resources, including a shortage of funds from government agencies, and an increasing number of referrals without sufficient clinic space.

- **Public and patient education:** The rapid increase in genetic information available; need for public education about the importance of knowing their family history and providing it to their health care providers; addressing public misconceptions about genetic services; and public education about the value of genetic services were all cited. Also, a need to address patient privacy concerns and fears about insurance and employment discrimination were discussed.

- **ELSI:** The influence of political and social climates on the availability of genetic services, stem cell research, pregnancy termination, and anxiety about “the Gattaca possibility”165, were among the concerns expressed by respondents. There were also ethical concerns raised regarding when and who to test, general population screening, direct-to-consumer marketing of genetic tests, constraints imposed by gene patenting, an increasing ability to screen for complex traits, and changes in molecular genetics and cytogenetics.

164 See [http://www.ahrq.gov/clinic/uspsfab.htm](http://www.ahrq.gov/clinic/uspsfab.htm). “The mission of the USPSTF is to evaluate the benefits of individual services based on age, gender, and risk factors for disease; make recommendations about which preventive services should be incorporated routinely into primary medical care and for which populations; and identify a research agenda for clinical preventive care.” Accessed September 20, 2005.

165 This refers to the movie Gattaca, which is a “science fiction drama, set in a future when one’s life is determined by genetic engineering rather than education or experience.” [http://www.blockbuster.com/catalog/DisplayMoreMovieProductDetails.action?BB=true&movieID=111966&channel=Movies&subChannel=sub#Full](http://www.blockbuster.com/catalog/DisplayMoreMovieProductDetails.action?BB=true&movieID=111966&channel=Movies&subChannel=sub#Full)
IDPH Supportive Efforts

A key question for this needs assessment project was: *What could the Illinois Department of Public Health do to better support you as a provider of genetic services?* The suggestions focused on funding, coverage/reimbursement, and education. There were also acknowledgements of IDPH’s current efforts, such as: “I have had no complaints over the years. I support the efforts [by IDPH] to become more involved in areas that affect a greater percentage of the general population (cancer, heart disease, etc.).”

- **Funding** – Needs included: more funding to support genetic counselors; grants for genetic counselors to obtain continuing education; grants for outreach and education programs; continued support for initiatives in genetics; and pleas to continue funding as well as advocate at the legislative level to ensure that funding is maintained;
- **Coverage/Reimbursement** – (Please note that some of the suggestions may not fall under the purview of IDPH.) Respondents suggested paying for cancer susceptibility testing, raising Medicaid reimbursement rates for physician services, recognizing genetic counselors as health care providers who can bill in their own right (and developing appropriate billing codes), and providing services to indigent populations. Respondents asked that IDPH lobby/work with Illinois insurance companies to cover more genetic services;
- **Education** – There were suggestions regarding education for primary care providers, other health care providers, the public, and genetic providers.
  - **Primary Care Physicians** – Provide programs at the local level, grand rounds, local conferences with follow-up interactive Internet-based learning tools, an “active awareness campaign of genetic services in Illinois,” education about available services and resources, support graduate education, provide information about what tests are considered standard of care;
  - **Public/Patients** – Create public service announcements/campaign about the importance of genetic counseling and availability of resources, and use public education and patient outreach to lessen the stigma of genetic diseases, patient outreach. As two respondents noted:
    - “IDPH focus should be patient education, patient education, patient education, beginning in the education-setting and the health setting – i.e., public health genetics starts with an educated populace.”
    - “I believe that simply supporting and promoting awareness of genetic conditions and genetic medicine will make our specialty more viable and provide us with the ability to support ourselves.”
  - **Genetic professionals** – Offer workshops on genetic counseling for common complex diseases, help counselors “educate the medical community about the potential of genetic services to prevent illness,” promote genetic provider services to non-genetics professionals, “offer education to bridge the gap between genetic research and clinical application;”
  - **Others** – Educate community nurses and hospitals about newborn screening and related services.
- **Other** – Gather data on the cost-effectiveness of genetic services, develop and implement screening tools to identify at-risk individuals, increase the number of medical geneticists in certain areas, help lobby for the Genetic Non-Discrimination Act, encourage collaboration with local health departments, and build better networks with national organizations, such as the March of Dimes. There was also a request to establish a “genetic services guidance committee – with open membership, including genetic counselors, which will consider
issues other than newborn screening.” Another request was for IDPH to re-evaluate the effectiveness of providing grants to county health departments.

Conclusions

This section provided demographic and practice data for genetic service providers in Illinois. It is important to note that the results from these survey respondents may not be generalizable to all genetic providers in Illinois. However, it is of interest that the results of this survey are consistent with the recently published results of a national survey of American Board of Medical Genetics (ABMG) certified geneticists (Cooksey et al., 2005).

Almost all genetic service providers were white and non-Hispanic. Over half of geneticists and all but two genetic counselors were women. The youngest geneticist respondent was 41 years of age, with 34% aged 55 years or older. In contrast, 60% of genetic counselors were less than 35 years of age, and almost half (47%) had been at their current place of employment for less than five years. On average, geneticists worked 51 hours per week, while genetic counselors worked 40 hours per week. Only 8% of respondents reported that their practice was full.

Eighty percent of respondents worked either in an academic medical center/university or a hospital. About half of respondents were comfortable with staffing levels at their primary work setting. Compared to medical geneticists, genetic counselors spent more of their time in clinical patient care, had a larger prenatal/reproductive patient and smaller infant/child caseload, and saw a larger percentage of managed care patients and fewer uninsured patients. Most providers discussed risks with patients when recommending susceptibility testing or presymptomatic testing. There was support for potential telemedicine use, but limited experience with it. Sixty-two percent of respondents reported being very familiar or familiar with IDPH’s Genetics and Newborn Screening Program.

Respondents provided a great amount of information in their open-ended responses. They expressed concern with insurance coverage and reimbursement issues, including constraints regarding payment for genetic services; lack of physician education regarding genetics; the size and distribution of the genetic provider workforce; and the need for public education. Many concerns and uncertainties were described regarding the field of genetics in five to ten years, notably questions about the roles and responsibilities of trained genetic subspecialists and other non-genetics health professionals. In general, respondents felt that IDPH should play a role in genetic services funding, coverage and reimbursement issues, and education.
Chapter 6: Interview Findings

Key informant interviews were conducted with several relevant groups of stakeholders: genetic service providers (including medical geneticists, laboratory geneticists, and genetic counselors), non-genetic health care providers (including primary care and specialty providers), local health department staff, state agency personnel, and representatives from consumer/advocacy organizations (for more detail about sample selection, the interview process, and response rates, see Appendix 1: Methods; for copies of the interview guides, see Appendix 4). The findings from these interviews are presented below based on stakeholder group.

Genetic Service Providers

Thirty four genetic service providers were interviewed, including: fourteen MD geneticists, two PhD geneticists, three MDs not certified in genetics but directly engaged in the provision of genetic services, fourteen genetic counselors, and one genetic nurse. Interviewees represented all sixteen Illinois regional genetic centers receiving funding from the Illinois Department of Public Health (IDPH), as well as other genetic services centers that do not receive IDPH funding. The IDPH-funded centers are located throughout the state and serve additional areas through outreach clinics. Unless otherwise noted, in this section the term providers refers to genetic service providers.

During the interviews, genetic service providers provided their perspective on the changing field of medical genetics, the challenges they face in the provision of care, and the key issues they see facing the state. They were asked to consider how effectively primary care providers are integrating genetics into patient care, identify needs for service provision and barriers to care in the state, and delineate key roles for the state department of public health in the genetic service system. The findings are presented below.

Praise for IDPH’s Genetics and Newborn Screening Program

In the interviews, a number of providers expressed appreciation for the work and accomplishments of IDPH’s Genetics and Newborn Screening Program. Some of this praise includes:

“We have a very strong department of public health [in Illinois]. . . . I am very pleased with our state being proactive.”

“IDPH has done a great job in terms of newborn screening.”

“We have a lot of really good professionals in the state who are really dedicated to improving genetic services.”

“I think that [IDPH] has done a good job in terms of the whole issue with expanded newborn screening. I feel proud of our state in that we have that available.”
The Changing Field of Medical Genetics

Genetic service providers were asked to describe how they see the field of medical genetics changing over the next five to ten years. The principal issue that participants identified (thirty of the thirty-four interviewees) was the rapid expansion - of technology, knowledge, service, and demand - in genetics. Specifically, providers suggested that expansion would be significant in:

- the number, range, availability, and complexity (both technical and ethical) of genetic tests (twenty),
- the clinical relevance of genetics to a wide range of common, chronic health conditions (nine) and to genetics-related treatment options (one), and
- the demand for services, given the increased direct-to-consumer marketing and availability of tests (two).

Representatives commented that genetics will include more common diseases and expressed concerns that the ethical issues are not keeping pace with the advances in technology. One participant summed up the sentiments of many: “I think [the field of genetics] is going to expand exponentially. There are going to be many more [genetic tests] in the coming years. The field is going to get far ahead of the knowledge base of most care providers.”

Providers also anticipated that the nature of consumer demand would change (due to direct-to-consumer marketing of genetic tests, for example, consumers may come to providers after obtaining test results), raising important practical and ethical issues. One participant expressed concern that considerations of access to testing not obscure similar considerations of access to treatment.

Emergent Models of Care in Medical Genetics

Considering the impact of this expansion on the practice of medical genetics, participants indicated that they expect to see:

- increasing provision of genetic services by non-geneticists, especially primary care providers (eleven);
- increasing disease specialization among geneticists and genetic counselors (three); and
- changes in the organization and nature of genetic counseling (three).

One interviewee felt that the ‘field of genetics’ as such would end, as responsibility for assessment and testing shifted increasingly to primary care providers and other specialists, and that genetic service providers would work in a more specialized capacity with patients in need of more complicated care. Another participant noted that education programs would increasingly adapt their training models toward greater specialization.

In addition, three providers suggested that the nature, role, and organization of genetic counseling may change. One suggested that genetic counselors would increasingly work in specialty areas with physicians who are not geneticists. Two participants suggested that the changing technologies may alter the way genetic counseling is conducted, with one saying that the expansion in testing options may precipitate a shift from counseling for specific tests, to a more general genetic counseling that would cover a range of tests.
Broad Implications of Expansion

Participants identified the following broad implications resulting from the rapid growth in the field of genetics:

- A need to ensure a genetics-competent primary care and specialist workforce (eleven):

  A clear concern about the knowledge deficits and educational needs of the greater health workforce emerged from the interviews. One provider noted that, given advances in treatments, early identification of patients by primary care providers would become crucial, saying “that means that primary care physicians need to be extremely well educated to get patients to us early. That means that they need to know much more than they know now about the early signs of genetic conditions.” While the majority of participants emphasized the need to increase the genetics knowledge of primary care providers, a few also raised questions about what primary care and other health care providers should know. This suggests that determining the nature of competencies will also be an important activity as genetics becomes increasingly relevant to mainstream health care.

- A need to consider the social and ethical implications of genetic technologies and care (seven):

  Three participants noted a need to address ethical, legal and social concerns, specifically insurance discrimination and consumer protection. Four indicated a need to address health disparities, which may be exacerbated or created in the context of expanding genetics care, and access constraints related to the cost of genetics care. One provider discussed cost and ethical issues related to the projected increase in the availability of genetics tests and asked: "How do you incorporate new information, how does it become standard of care, and how do we pay for it?"

- A need to define professional roles of genetics and non-genetics health care providers and to assure quality of care (six):

  Six providers anticipated that confusion about professional boundaries and quality assurance in the provision of genetic services would demand greater attention. One participant expressed concern about this confusion and its impact on quality as more and more health care professionals become ‘genetic providers,’ offering similar services but with greatly differing levels of knowledge. Another interviewee referred to current professional divisions between laboratory geneticists (PhDs and MDs certified in genetics) and pathologists, and asked who should be considered professionally qualified to analyze specialized genetic tests. He suggested that such divisions were likely to “snowball” in the coming years. Representative comments included:

    Genetics is going to play a role in so many other areas. So many areas are not ready for that to come. It is going to require that more people get trained or more people know how to make appropriate referrals.

    “Who will be on the front lines delivering the services? Genetic counselors or family practice physicians? . . . .What will be our [genetic services provider] role?”

- A need to articulate clear standards of care responsive to increasing genetic knowledge and technology (three):
Three participants suggested that a key issue facing the field of genetics, and health care more generally, will be determining how to incorporate the rapidly expanding body of knowledge about genetics into effective and appropriate standards of care.

What’s going to happen is there will be increased competition where people who do not have the training, the expertise, the experience in genetics will be providing these kinds of services because there are no guidelines, there are no boundaries... there has to be some kind of balance between who does the genetic services and maintaining an individual's perspective on what these services really mean.

Challenges Facing Genetic Service Providers and the State

Genetic service providers were asked to describe what they perceived as: (1) the major challenges to their provision of genetic services; and (2) the key issues the state will face in the coming five years. There was substantial overlap in the responses to these two questions. In the following section, provider responses to both questions are summarized (Table 6.1).

<table>
<thead>
<tr>
<th>Issue identified as…</th>
<th>key issues facing the State number responding=</th>
<th>a challenge to providing care number responding=</th>
</tr>
</thead>
<tbody>
<tr>
<td>Finance/Reimbursement</td>
<td>22</td>
<td>20</td>
</tr>
<tr>
<td>Workforce</td>
<td>10</td>
<td>12</td>
</tr>
<tr>
<td>Education</td>
<td>8</td>
<td>10</td>
</tr>
<tr>
<td>Access</td>
<td>8</td>
<td>3</td>
</tr>
<tr>
<td>State support for genetic services</td>
<td>9</td>
<td>0</td>
</tr>
<tr>
<td>Insurance/cost</td>
<td>0</td>
<td>6</td>
</tr>
</tbody>
</table>

Finance/Reimbursement

Twenty participants identified inadequacies in the current systems of finance and reimbursement for genetic services as a key challenge to providing service, and twenty-two identified this as a primary issue facing the state (Table 6.2).

Table 6.2 Participant Responses Regarding Finance/Reimbursement Issues

<table>
<thead>
<tr>
<th>Issue identified as…</th>
<th>key issues facing the State number responding=</th>
<th>a challenge to providing care number responding=</th>
</tr>
</thead>
<tbody>
<tr>
<td>Finance/Reimbursement</td>
<td>22</td>
<td>20</td>
</tr>
<tr>
<td>Inadequate 3rd party reimbursement for genetic testing/services</td>
<td>17</td>
<td>11</td>
</tr>
<tr>
<td>Inadequate Medicaid reimbursement for genetic testing/services</td>
<td>12</td>
<td>8</td>
</tr>
<tr>
<td>No reimbursement for genetic counseling services</td>
<td>1</td>
<td>5</td>
</tr>
</tbody>
</table>
The most frequently specified challenges/issues were inadequate third party reimbursement policies for genetic testing and services, with an emphasis on Medicaid, and the absence of mechanisms enabling reimbursement for essential components of care, such as genetic counseling and intellectual time. Explaining the difficulties departments and providers face negotiating coverage with third party payers, one interviewee discussed how the institution ends up absorbing the costs when insurance will not cover services and the family will not or can not pay; he noted the need to meet a certain standard of care, irrespective of insurance coverage.

Participants also noted that they faced challenges with respect to the time demands of negotiating coverage with third party payers on a patient-by-patient basis. Expressing common frustrations, one interviewee noted, in regard to getting genetic services for needy patients and getting the appropriate testing paid for: “I didn't take a course in buzzwords in medical school to write letters [of medical necessity]... It used to be a miniscule part of what I do and now it is a major part of what I do, talking to insurance companies and writing letters...And it takes a lot away from the clinical part.”

Medicaid was the focal point for the majority of providers who identified reimbursement as an issue. Most indicated that Medicaid covered a large or disproportionate number of their patients, and that restrictive Medicaid policies limited access to care for patients and often shifted the expense to genetics providers and/or departments. One participant commented that the Medicaid program was more restrictive and rigid in its refusal to cover services than private insurance companies. Commenting on the disparities this can create, this provider described “losing” a lot of African-American patients who need hereditary breast/ovarian cancer genetic testing because they are on Medicaid, which does these screenings; this provider also indicated that while private insurance companies usually “come around” and provide coverage, Medicaid does not.

The lack of a mechanism enabling genetic counselors to bill directly for their services was identified as a key challenge by five participants and as a major issue facing the state by one. Both medical geneticists and genetic counselors identified this as an issue. Two interviewees specified that insufficient funding from the state for genetic counseling compounded the difficulty of providing this service. Providers explained that because genetics departments in hospitals and medical centers are unable to bill third party payers directly for genetic counseling services, financing these services requires utilizing state resources and revenue generated from other clinical services. In some genetics departments, a significant proportion of the cost of genetic counseling is financed through the state clinical genetics grant program. The struggle to financially cover a non-reimbursable part of what providers consider an invaluable component of genetics services creates an ongoing challenge. Commenting on this, one interviewee noted:

Whatever is not covered by the grant...my Department has to pay for... I get the money to pay for that from clinical income from other providers, physicians... I personally think [genetic counseling services] are an important thing for us to have but it is problematic... We have two full time genetic counselors... and could probably fill up a third person but can't afford it... But how do you bill for them?... How do they get reimbursed?

In addition to reimbursement for counseling services, two interviewees identified the absence of reimbursement mechanisms for intellectual time - time spent researching diagnoses and care- as a key challenge to providing care. As one provider noted:

My procedure [for coming up with a diagnosis] if I don’t recognize [a syndrome] is to go to the genetic databases. I might spend two to three hours searching…. and I can't get paid for that... I might be able to charge for it but I won't get reimbursed for it. I spent as many years
in training as any cardio-thoracic surgeon, but what I do is not valued. That's my procedure. It’s an intellectual thing, but we don't get paid for intellectual time. There are no billing codes. And it's not face-to-face.

Other finance/reimbursement issues included low salaries for MD geneticists; the inability, given the difficulties with reimbursement, for geneticists to generate enough revenue to support themselves institutionally; and a lack of institutional support for genetics.

Workforce

Workforce issues were the second most frequently cited challenges facing providers (twelve) and the state (ten; Table 6.3). In addition to common concerns about the total number of current and future providers, two participants noted that many currently practicing geneticists will be retiring in the next few years, and that not enough candidates are entering the field to ensure adequate replacement levels. One interviewee commented, in the context of increasing availability of genetic services, “We are going to need dozens, if not hundreds, more genetic counselors around the state. . . . [E]ven if you combine all the graduate programs in genetic counseling, we're not going to be seeing sufficient numbers of genetic counselors for decades to come.” One participant noted that this was of particular concern in rural areas, which are already underserved due to the uneven distribution of genetics professionals in the state.

Table 6.3 Participant Responses Regarding Workforce Issues

<table>
<thead>
<tr>
<th>Issue identified as…</th>
<th>key issues facing the State number responding=</th>
<th>a challenge to providing care number responding=</th>
</tr>
</thead>
<tbody>
<tr>
<td>Workforce</td>
<td>10</td>
<td>12</td>
</tr>
<tr>
<td>Inadequate supply of genetics providers</td>
<td>5</td>
<td>4</td>
</tr>
<tr>
<td>Inadequate clinical time to meet demand</td>
<td>0</td>
<td>6</td>
</tr>
<tr>
<td>Ineffective incorporation of genetics into primary care practices</td>
<td>3</td>
<td>2</td>
</tr>
<tr>
<td>Licensure of genetic counselors</td>
<td>4</td>
<td>0</td>
</tr>
</tbody>
</table>

A number of interviewees expressed concern about the ability of current genetics providers to meet the expected increase in demand for services. Six participants suggested that this was a reflection of inadequate staffing and finance/reimbursement systems. Another shared her frustrations with trying to provide the highest standard of care in a context of increasing time constraints and demands.

Three participants identified primary care providers’ lack of incorporation of genetic information into their provision of care as a major issue facing the state, and two viewed this as a challenge to providing care. Specific concerns included primary care providers’ inability to provide effective counseling on genetic screening options; the inappropriate management of patients with genetic conditions; and the failure to offer standard prenatal screens. As one participant noted: “If I am not around, I worry that patient care is not given on a timely basis or in an appropriate service.” (This issue will be explored further in the section entitled, “Genetics and Primary Care Providers.”)

Four participants discussed the recent passage of legislation requiring licensure for genetic counselors as a major issue, suggesting that licensure may facilitate the establishment of
mechanisms enabling genetic counselors to receive reimbursement for their services. They also suggested that licensure may affect the development of future models of care in genetic service provision by, for example, enabling genetic counselors to practice independently.

Two genetics professionals expressed concern that, as a group, they were being underutilized by the larger health care system in the state, with one observing that a lack of clear professional boundaries is developing between themselves and other specialists. One interviewee indicated the need for more support staff in genetics divisions at medical centers, and another noted an inadequate number of laboratory personnel as a key challenge. One participant also raised the issues of negotiating the legal liability of expanding prenatal testing options.

Education

Ten participants identified a lack of education about or awareness of genetics among other health care providers and the public as key challenges in providing genetic services, and eight identified the same as key issues facing the state.

The majority of responses to both questions focused on the educational needs of primary care physicians and other non-genetics specialists and sub-specialists. Interviewees found that these providers lack knowledge/awareness of genetics and seem to perceive that genetics applies only in the case of rare diseases, contributing to their often ineffective identification of genetic risks and referral of patients. These knowledge deficits were believed to contribute to inappropriate utilization of tests, misinterpretation, and inappropriate patient care. Expressing how this impacts her practice, one provider commented: “Other care providers do not see the beginning nor the end of genetic testing. They do not know when to begin and they do not know what its limits are... I am so challenged with that because that means that I need to educate them.”

These participants suggested that a key issue facing the state will be the educational needs of the primary care workforce and the medical system more generally. Interviewees noted that it will be crucial in the coming five years to ensure that health care providers are educated enough to effectively negotiate and incorporate genetics information into patient care. Commenting on the timeline for this, one participant stated that another key issue is “educating the primary care physicians about genetics and genomics and why it's important in their practice now and not in ten years.”

Providers also noted issues related to public perception and knowledge of genetics. These included: public misperceptions about what genetics can and cannot do, a general lack of awareness about genetic services and options, public conflation of genetics with abortion, and public fears that genetics means something ‘bad.’

Access

Eight participants identified access to care as a key issue facing the state and two as a challenge to providing care. The most frequently noted concern was the uneven distribution of genetics providers and services throughout the state. Participants explained that the majority of providers are currently concentrated in Chicago and a few other urban areas, leaving large areas of the state un- or underserved, especially in the southern portion of the state. As one interviewee noted, the challenge is:

…figuring out ways to reach [people] who don’t have access to even traditional genetic services...There are maybe 70 genetic counselors in Illinois and probably 50 of us work in Chicago... There is a large portion of the state that is not receiving even traditional genetic services. That remains a challenge and it's one we have not been focusing on.
In addition to the uneven distribution of services, two participants identified a related concern - inadequate transportation options for patients in non-urban areas. One interviewee included the problem of language barriers, specifically a dearth of providers with facility in languages other than English.

State Support

Nine participants indicated that a key issue facing the state over the next few years will be the degree of the state’s commitment to genetic services, including the continued provision of its newborn screening and genetics programs. All nine interviewees expressed concerns that state programs and financing for genetic services might be cut, either directly or indirectly (through cuts to Medicaid), which would reduce the viability of service provision to a significant portion of the state’s population. Two participants indicated that their concerns were shaped by what they perceived as a lack of understanding or appreciation at the state level for the value of genetics; one referred specifically to the recent state-directed transfer of money from the newborn screening fund to the general revenue fund as evidence of this, commenting:

I am very concerned about . . . the budget slashing and current [poor] Medicaid reimbursement rates... and I think there is a tremendous threat to health care in general. I am concerned about the fact that money was slashed from the newborn screening budget a year ago. I am concerned that could happen again. In a time when we need to be investing more in funding for genetic health care programs, I am concerned that the climate suggests that just the opposite is going to happen . . . . We are behind the times... in terms of recognizing that we need to spend money on our state's children. Anything else would be a far distant number two.

Insurance/Cost

Six interviewees discussed the issue of insurance coverage and cost as key challenges they and their patients face in providing and receiving genetics services. In particular, they were concerned about the expense of services for the patient or the effect of the limitations of coverage on a patient’s decision to utilize services. Two of these participants noted the challenges faced caring for patients without insurance, for whom the costs of testing can be prohibitive. Four providers discussed the ways in which restrictions and limitations imposed by insurance companies directly limit patient care. One provider explained that it is often difficult to obtain insurance coverage for tests that may seem “esoteric,” are “expensive,” or if the laboratory used is out-of-network (even though it may be the only laboratory performing a specific test). This participant continued by expressing frustration that insurance companies do not seem to understand that comprehensive genetic care includes both genetic counseling and genetic testing and, therefore, many patients are not receiving both services.

Noting how this affects patients’ decision-making processes, another provider stated:

We have noticed more and more patients that have called to cancel genetic counseling because it is not a covered service through their [insurance] provider and they are not willing to pay out-of-pocket for what they feel is an unnecessary service. They want the ultrasound or the amniocentesis... [We will do the ultrasound without genetic counseling, but not the amniocentesis] . . . [Lack of insurance coverage] is a huge stumbling block.
One interviewee shared that, in addition to issues of service coverage, it seemed that many patients were not receiving services for fear of insurance discrimination. This provider commented, “Many times patients don’t come here because they are afraid their insurance is not going to cover it or that just by coming they will be red-flagged [by their insurance] as far as being at increased risk. That is one of the major challenges.”

Genetics and Primary Care Providers

Genetic service providers were asked to describe whether, in their experience, primary care providers were effectively incorporating genetic information into their practices. They were also asked to comment on how primary care and genetic service providers might better complement each other.

Although the interviewers supplied no definition of ‘primary care provider,’ genetic service providers included the following types of physicians in their responses: obstetricians, gynecologists, pediatricians, family practice physicians, and general practitioners.

The term ‘effectively incorporating genetic information’ was not defined by the researchers but was derived from genetic service providers’ responses. Fourteen interview participants identified both – and two identified one or the other - of the following as ‘effective incorporation’ of genetics information:

- to evaluate for or recognize genetic risk factors (taking family histories and/or considering the role of genetics in evaluating their patients); and,
- making effective referrals for patients who might benefit from genetic counseling or evaluation.

A representative comment was that:

The primary care provider's role is very important, because most people see [them] much more often than they see a specialist. So if they just take the time to ask about the family history and ask about [the patient’s] concerns, it might just open the door for the patient that might not bring it up on their own.

Incorporating Genetics into Primary Care

Seventeen participants provided mixed assessments of primary care providers (PCPs) in response to the question of whether they are effectively incorporating genetic information into their practices. Thirteen interviewees noted that some PCPs do better than others; four suggested that some do better with certain conditions (e.g., Down Syndrome) or activities (e.g., referral); and two commented that PCPs were incorporating genetics information to only a limited extent. Three participants indicated that there had been improvement in PCP’s facility with genetics in recent years.

Thirteen genetic service providers stated that PCPs are not effectively incorporating genetics into their practices (twelve) or that only a very few are (one). Three interviewees were unable to

---

166 Note that two providers provided more than one assessment, for example noting that some are doing better than others and that they are doing better with some conditions than others. Thus, the count for mixed assessment is two over the actual number of providers that provided such an assessment.
answer the questions, and one replied that, in her estimation, primary care providers were effectively incorporating genetics into their practices.

Genetic service providers identified a number of areas where PCPs were failing to effectively incorporate genetic information. Four participants stated that primary care providers are not aware of genetics or of how it might be of value to their patients. Another four interviewees each noted that PCPs: did not possess and/or were not sharing accurate genetic information with their patients; were not making appropriate clinical recommendations to patients with genetic conditions or risks; or were not providing appropriate counseling to patients regarding screening options. One participant stated that some patients had been given information that was "way off in left field.”

Four providers said that PCPs were failing to offer standard genetic screening tests despite patient indicators (e.g., failing to offer cystic fibrosis screening to Caucasian patients), or, as one noted, failing to order specialty genetic tests. Five participants indicated that PCPs were not able to accurately interpret the results of genetic tests, including the results of the state’s mandated newborn screening tests.

Constraints in Incorporating Genetics into Primary Care

Nineteen interviewees reflected that primary care providers do not have sufficient education, knowledge, skills, or experience to effectively assess, refer, or counsel patients appropriately on genetic testing options and outcomes.

Specific knowledge deficits they identified include:

- how, when, and where to make appropriate referrals (noted by six participants);
- genetic conditions, risk factors, and indications (five);
- basic genetics, such as how genes work (three);
- indicators for genetic tests and/or of test interpretation (three);
- patient options regarding genetic screening (two);
- skills to effectively counsel patients on testing options (two); and,
- the management and appropriate care of patients with genetic conditions (two).

Seven participants identified a lack of time – for both continuing medical education in genetics and for adding genetics to the clinical patient visit - as a key constraint on PCPs. Three providers recognized the difficulty of carving time out of a clinical practice to participate in genetics-related educational activities. Six interviewees noted that PCPs simply do not have the clinical time to either conduct the necessary background research or provide the comprehensive counseling that is crucial to genetics care. Revisiting the issue of non-reimbursable activities, such as research and counseling, one provider noted:

One of the problems is, as genetic information and genetic testing become more and more available, the whole issue of who has the time and who gets reimbursed to explain clearly these options because decisions [and ramifications] to or not to take a genetic test are not always that simple... [This] is a serious problem.

Elaborating on the lack of adequate clinical time, one interviewee noted that patient counseling includes a discussion of costs, insurance, test interpretation, and ethics to help patients make decisions. This provider elaborated: “A person who is unaffected and tested without any counseling, that is when genetics should come into play. Primary care physicians typically do not go into the kind of detail that needs to be conducted in those sessions.”
For one interviewee, the convergence of constraints on time and knowledge informed her perspective that primary care providers should not be “doing genetics.” Noting the enormous amount of time he/she spends on each patient, this provider said:

We are expecting primary care physicians to do more and more management of increasingly complicated patients. This field that is changing every day is just too much to ask somebody to try and do. I think geneticists have made an enormous mistake by saying that the primary care physicians should be taught to do [genetics], because there isn’t any way to teach the primary care physician to do it.

It is of interest to note that while all types of providers interviewed identified knowledge and educational needs as key issues, the issue of time constraints for physicians was only identified by the interviewees who were medical doctors.

Defining a Role for Primary Care in Genetics

Genetic service providers were not asked to define the role of primary care providers in the provision of genetic services. However, we assessed genetic service providers’ explicit statements on the role of primary care providers and considered the emphasis placed by interviewees on the tasks/activities that primary care providers fail to perform. Many participants (fifteen) consider the principal role of primary care to be risk or disease recognition and referral. Three providers specifically stated that the ideal role for PCPs is in recognizing risks for genetic conditions and referring patients as indicated to genetics professionals. Both education and time constraints were understood to affect the capacity of primary care providers to fulfill these roles, with one participant noting, “The biggest issue is just education. In my experience, and I think other studies have shown, the more educated somebody is about different genetic conditions, the more likely they are to refer.”

Areas of Improvement

Seven providers indicated that they felt that primary care physicians were successful at disease recognition and referral, although some suggested this varied by condition, with some more readily recognized than others. Three interviewees noted that they had seen recent improvement in PCPs’ ability to refer or manage patients with genetic conditions, and two providers indicated that they expected to see greater integration in the coming years. Further, as mentioned above, one provider responded that she felt primary care providers were effectively incorporating genetics, based on the frequency and appropriateness of referrals she receives and the types of tests they order.

Collaborative Possibilities

When asked how the roles of primary care physicians and genetic providers can better complement each other, twelve participants responded with the following suggestions:

- increase primary care providers understanding of genetics,
- improve communication, especially with respect to individual patient care, and
- develop the referral relationship as a more effective process of collaboration.
Reflecting on this question, one provider noted that there is often an assumption among geneticists that primary care physicians need to change to accommodate the emerging field of genetics. A genetic service provider suggested that, conversely, geneticists need to learn how genetics can best fit within their (PCP) models of care, stating that “We need to figure out how we can work [genetics] into their system rather than walking in the door and saying, ‘This is how we do it so you have to do it this way.’ I think that has been a big barrier in trying to merge with some of these other specialties because genetic systems are so different.”

Some genetics providers were careful to note that they did not expect primary care providers to become, in the words of one provider, “mini-specialists.” A few interviewees recognized as unrealistic the idea that primary care providers would independently manage patients with complex genetic conditions.

### Barriers to Accessing Care

Genetic service providers were asked to describe what they see as the primary barriers to patients’ access to care in Illinois and identified the following (Table 6.4):

**Table 6.4 Barriers to Accessing Patient Care for Genetics Services**

<table>
<thead>
<tr>
<th>Barrier</th>
<th>Number Responding</th>
</tr>
</thead>
<tbody>
<tr>
<td>Uneven geographic distribution of genetic services providers</td>
<td>14</td>
</tr>
<tr>
<td>Too few providers around the state</td>
<td>9</td>
</tr>
<tr>
<td>Lack of education, awareness, appreciation of genetics among PCPs</td>
<td>13</td>
</tr>
<tr>
<td>….among the public</td>
<td>11</td>
</tr>
<tr>
<td>Expense of genetic services and testing</td>
<td>8</td>
</tr>
<tr>
<td>Inadequate number of genetics providers</td>
<td>6</td>
</tr>
<tr>
<td>Lack of/ineffective genetic services at the primary care level</td>
<td>5</td>
</tr>
<tr>
<td>Lack of cultural and linguistic diversity among genetic service providers</td>
<td>2</td>
</tr>
<tr>
<td>Negative public perceptions of genetics</td>
<td>2</td>
</tr>
<tr>
<td>Conservative social and political climate</td>
<td>2</td>
</tr>
</tbody>
</table>

- An uneven distribution of genetic services providers, reflected in a concentration of services in Chicago and a few other urban areas (fourteen), and
- A dearth of providers and services throughout most of the state (nine).

Access to transportation, travel time, and travel costs were also noted as key barriers. One provider suggested that IDPH focus its resources on assuring service provision in the underserved areas of the state. Another noted that many patients from non-urban or smaller urban areas express discomfort at the thought of traveling to Chicago for services. This participant noted that yet another barrier:

…is the reliance on [educational institutions] to see patients.... The specialists kind of gather around the large educational institutions, medical centers. . . . As long as the state
[relies] on these large medical centers they are not going to reach people. I think if you had to go to a super-specialist... a person from northern Illinois would go to Madison or Iowa, not to downtown Chicago. . . . The attitude is, ‘Let the patients come to us,’ and that don’t work.

- Lack of education about, awareness of, and appreciation for genetics among primary care providers (thirteen) and the public (eleven).

Thirteen providers noted a general lack of education and awareness about genetics among primary care providers. In addition, some genetic providers suggested that primary care providers did not seem to value genetics or understand how it might contribute productively to patient care. On this point, one provider noted:

I think that there's a general lack of understanding about the value of genetic services that's both with patients and with physicians. There are physicians who will downplay a patient’s request for genetic services... and there are patients who are referred and they just don't understand what it's all about and don't want to bother.

Similar concerns were expressed with respect to level of education/awareness among the public. Eleven participants felt the public lacks knowledge about services and options, and how genetics can play a role in health care, and identified this as an important barrier to patients’ utilization of services.

- Inadequate funding, reimbursement, and/or insurance coverage for genetic services (eleven).

Eleven interviewees noted a lack of stable and sufficient funding for genetic services and inadequate third party reimbursement as barriers to access to care. Again, Medicaid restrictions were specifically mentioned. As one provider put it, “Medicaid doesn’t pay for any DNA testing. What that means is that it's not a service available to people covered by Medicaid. In Illinois, I understand that is [a significant] percent of newborns.” The absence of insurance coverage was also noted as a barrier, and one participant expressed the view that insurance coverage may be shaping which referrals are being made.

- The expense of genetic services and testing (eight).

Eight providers noted that the expense of genetic services was a key barrier for patients who must pay out-of-pocket. One expressed concerns about how the expense may be affecting disparities in service provision, noting:

Similar to other medical services where you end up seeing educational and financial barriers to genetic services, I often don't feel that genetic services are offered at the same level to people of lower income or lower education as they are to other groups. . . . [We need to] bridge the gap to make sure that genetic services are being offered uniformly to all groups.

- Inadequate number of genetics providers (six).

Six interviewees expressed concern that an inadequate supply of genetic providers was creating barriers to patient access. One provider commented that if all appropriate referrals were currently being made, there would be an immediate shortage of genetic counselors and geneticists in the state.
• Lack of effective genetic services at the primary care level (recognizing risks, making referrals, and offering standard or specialty genetic tests) (five).

Five providers noted that a key barrier to patients’ service utilization is primary care providers’ lack of effective integration of genetics into their clinical practices. This affects both what services primary care providers are offering to patients and the degree to which primary care providers are making appropriate genetic referrals for those patients who might benefit from them. Expressing concern about the degree to which primary care providers’ lack of awareness may be affecting patient access, one participant noted that patients usually see a specialist when their primary care physician recognizes a potential or actual problem and, with genetics, that knowledge base is lacking.

• A lack of cultural and linguistic diversity among genetics providers (two).

Two participants reported that linguistic and cultural homogeneity among providers may limit both access to services and the quality of care for patients with limited fluency in English. It was also noted that a lack of cultural awareness and sensitivity reduces the likelihood that genetic services providers are communicating effectively with patients of different ethnic/cultural backgrounds. One provider noted that, given the complexity of genetic topics, it is extremely difficult to conduct genetic counseling through a translator and all the more important to teach genetics providers to be culturally sensitive.

• Public perceptions that genetics is something frightening or bad (two).

Two providers commented on what they believe is a public perception that genetics is something frightening or bad, which they feel discourages utilization of services. Commenting on this, one participant noted:

I . . . think there is also a certain prejudice against genetics. There is a certain feeling that genetics is somehow - evil is probably too strong a term but maybe tampering with nature... There is just so much hype in the media about genetics in general, and prenatal testing, of course, is very controversial.

• Conservative social and political climate (two).

Two interviewees identified a conservative social and political climate as a barrier to care in Illinois. One suggested that more conservative providers might not be offering genetic services or information to patients. The other felt that a conservative legislature had made it difficult to get state support for genetic services, especially prenatal screening and genetic testing.
Services or Resources of Greatest Need

Genetic service providers were asked to describe the genetic services and resources they feel are in greatest need in the state and provided the following (Table 6.5):

**Table 6.5 Resources & Services Most Needed in Illinois**

<table>
<thead>
<tr>
<th>Need</th>
<th>Number responding</th>
</tr>
</thead>
<tbody>
<tr>
<td>Address geographic access disparities</td>
<td>9</td>
</tr>
<tr>
<td>Improve financing/reimbursement of services</td>
<td>9</td>
</tr>
<tr>
<td>Improve public’s genetic literacy</td>
<td>9</td>
</tr>
<tr>
<td>Improve other providers’ genetic literacy</td>
<td>8</td>
</tr>
<tr>
<td>Increase adult &amp; prenatal services</td>
<td>7</td>
</tr>
<tr>
<td>Improve communication/foster systemic integration</td>
<td>6</td>
</tr>
<tr>
<td>Address supply of genetics providers</td>
<td>4</td>
</tr>
<tr>
<td>Improve insurance coverage</td>
<td>3</td>
</tr>
</tbody>
</table>

- A need to address access disparities in non-urban areas (nine).

Nine providers noted a need to address the uneven geographic distribution of genetic providers and services in the state. Five noted a need for more genetic providers and services outside Chicago, especially in the southern part of the state. Two participants identified a need to address the transportation barriers clients face traveling to services from underserved areas of the state. Two providers suggested strategies for addressing the issue, including the utilization of telemedicine and the establishment of regional genetic centers. One also commented on divisions between urban and non-urban clinicians as an issue in this context, noting: “The big centers look down their noses at small town docs. That is a serious problem.”

- A need to improve financing of and reimbursement for genetic services (nine).

Nine providers identified a need to improve current systems of financing and reimbursement for genetic services in Illinois. Five indicated a need to establish a better and more stable system for funding genetic services. Two participants each noted a need to enable independent billing for genetic counseling services and for more resources from the state to ensure that access to care is not limited by patient ability to pay. One provider each noted a need for more resources from the state to expand genetic services into adult and specialty care and a need for resources to assist families with the provision of long-term therapeutic and supportive care for family members with genetic conditions.

- A need to improve genetic literacy among the public (nine).

Nine providers noted a need for public education campaigns to improve population level awareness and knowledge of genetics, including knowledge of basic genetics, awareness of the value and role of genetics in medical care, and understanding of what services are available and how to access them. To meet these needs, one suggested the development of regional centers for information dissemination, and another provider suggested incorporating genetics into high school level science curriculums.
• A need to improve genetic literacy and efficacy among primary care providers (eight).

Eight interviewees noted a need to improve primary care providers’ knowledge, understanding and facility with genetics, including knowledge of basic genetics, genetics-related risk factors and indicators, and about the referral process. One also noted a general need to educate all clinicians providing services at a “grassroots” level, and one provider explicitly called for the incorporation of genetics into nursing curricula at the BA level.

• A need for more adult and prenatal genetic services (seven).

Seven participants identified the need for expanded service provision in the area of prenatal and adult genetics. Four of the seven noted a need for more adult genetic services in the state, with one specifically emphasizing a need for services for cancer genetics, and the other three noting other adult genetic conditions (e.g., adult neurofibromatosis). The remaining three providers suggested that there is a need for more services in prenatal genetics, including expanded carrier and newborn screening.

• A need to improve communication and foster systemic integration among clinical genetics and primary and specialty care (six).

Six participants indicated a need for systemic integration of genetics with primary and specialty systems of medical care. These providers expressed the hope that increased communication and integration among these different systems of care might facilitate more effective utilization of existing genetic services. Commenting on the need to make genetics a part of mainstream medicine, one noted that genetic service providers need “to be seen as the specialists that we are... not an exotic referral, but something that is part of medicine.” Considering what a more effective model of genetic counseling might look like, one provider stated, “right now, what happens is the genetic counselor works in concert with the physician and the physician bills, but that's not the most efficient use of time. It might be more practical to have [genetic counselors] see patients on their own, be supervised, but they could bill themselves.” One interviewee also noted a need to incorporate genetics into nursing education and practice.

• A need for more genetic providers, including MD geneticists and genetic counselors (four).

With respect to the number of providers, three interviewees noted a need for more MD geneticists, two noted a need for more genetic counselors, and one noted a need for more clinicians trained in genetics.

• A need to improve insurance coverage and address implications of insurance discrimination (three).

Two providers noted a need to address the issue of insufficient and disparate insurance policies with respect to genetic services, and one identified a need to address issues related to insurance discrimination.
Role of the State Department of Public Health

Genetic providers were asked to describe what they see as the role of the state department of public health within the genetic service system in Illinois. Participants identified four key roles for the state:

- assuring an educated health workforce and informed public (nineteen);
- providing direct services (sixteen);
- assuring access to care (seven); and,
- addressing the inadequacies in the financing and reimbursement of genetic care (four).

Education

The majority of participants (nineteen) emphasized education as a key role for the state. Seventeen indicated that the state should do more to educate primary care providers and other non-genetic health care providers on appropriate identification, assessment, and referral for genetic services, and on appropriate use of genetic tests. It was also suggested that the state address primary care providers’ knowledge deficits with respect to the newborn screening program. One interviewee each suggested the state: expand its educational initiatives to reach a wider audience of health care providers; develop information sheets to assist providers in identifying and referring patients; establish conferences around the state to reach providers; and, establish a mobile presentation for providers on when, where, and how to refer a patient for genetic services.

Ten providers indicated that the state should be engaging in genetics-related public education campaigns with the goal of assuring that residents in Illinois are aware of genetics, how it might impact their health, and where and how they might receive services. One participant suggested that these efforts be targeted to non-urban areas, where there are few, if any, genetic service providers. Two suggested that the state focus on the development of brochures and fact sheets, which could be disseminated by community clinics and primary care offices. One suggested that the state’s educational initiatives to health care providers and the public emphasize the value of genetics over and above any specific content. This provider suggested that in addition to knowledge deficits, there was a crucial lack of appreciation for why and/or how genetics might be valuable to health among both health care providers and the public.

Direct Service Provision

Sixteen providers identified direct service provision as a key role for the state. The majority indicated that the state should maintain its current newborn screening (five) and clinical genetics (nine) programs. A number of others expressed concern that funding for these programs might be reduced. As one noted, without the support of the state a number of centers would not be able to provide services, which would significantly affect Illinoisans’ access to care.

While emphasizing the need to maintain these programs, three providers suggested that the state evaluate the effectiveness of its local health department and clinical genetics grant programs. Two of these providers expressed concern about whether the grant monies were being effectively utilized and whether patient identification and referral was occurring at an optimal level. One suggested that increasing the state’s level of investment in outreach, by increasing the number of satellite clinics offered through local health departments, might be an effective strategy for reaching patients in need. Another recommended that the state take measures to ensure grant monies are subsidizing services for public aid patients, and not being utilized to subsidize the services of people with private insurance.
Three providers suggested that the state expand its clinical genetics program to support the services of more genetic counselors and personnel, and two suggested that the state expand its genetics programs to more fully cover chronic diseases. One each suggested that the state: improve its ability to identify and refer children with hearing loss; increase support for follow up care of children identified with genetic conditions through the newborn screening program; and, provide support for interpreters to aid in the provision of services to non-English speaking clients. Though the majority of providers argued for the state’s involvement in the provision of genetic services, one participant explicitly argued that the state should not be investing in service provision, including the operation of a metabolic laboratory, but rather should be focusing on education and regulation.

Access

Seven interviewees identified ensuring equal access to care for individuals in non-urban areas and traditionally underserved communities in urban areas as a key role for the state. Five of the seven specifically noted that the state should address the uneven distribution of genetic services in Illinois. Four participants suggested the state increase resources to underserved areas by increasing the number of outreach clinics it funds along with the number of providers serving in those clinics. One suggested the state also address the transportation needs of patients in these areas.

One participant suggested that the state invest in addressing issues of access in low-income communities, communities of color, and new immigrant communities in Chicago and other urban areas. This provider also suggested that the state monitor and address current and emergent health disparities in genetics.

Finance/Reimbursement

Four participants suggested the state should be addressing the issues of inadequate reimbursement and financing for genetic services in Illinois. Three of the four suggested that IDPH work at either the state or national level to improve third-party payer reimbursement policies for genetic services. One suggested that the state work specifically to change and improve Medicaid coverage and policies.

Conclusion

Genetic service providers identified a number of key issues affecting the current and future provision of genetic services in Illinois, including what they perceive to be key needs and barriers with respect to care. These issues, which were identified repeatedly throughout the interviews, fall readily into the categories of finance/reimbursement, workforce adequacy, education, engagement, access, and a cluster of issues that have come to be known as ethical, legal, social issues or implications (ELSI). To summarize, from the perspective of genetic service providers, the issues that need to be addressed in the state include:

1) An inadequate and uneven system of third party reimbursement for genetic testing and clinical services.

   Genetic service providers called attention to the inequities in care created by the current systems of finance/reimbursement for genetic services. The result of such inequities is that services are rendered financially insecure and dependent on subsidy, and the costs are subsequently imposed on providers and departments. This affects both who can access services and the quality and extent of services that are provided (where, for example, testing
is covered but counseling is not). In the context of genetic services where the effects of testing may include long-term social consequences (e.g., genetic discrimination affecting insurance premiums or employment) and/or personal consequences (anxiety, depression, disrupted family relationships, etc.), the way insurance coverage policies limit the ability of providers to counsel patients is an important concern. Many providers expressed special concern for the limited services available to patients covered by Medicaid. It is clear from the interviews that these insufficiencies, and their consequences for providers and patients, are of paramount concern to genetic service providers. In addition to current effects, a few providers suggested that these issues may make the field of medical genetics less attractive to new medical students and residents. Lower salaries and the constant strain of fiscal instability are expected to impact the future adequacy of the workforce at a time when genetics is becoming an increasingly important aspect of health care.

2) The discrepancies between what are considered essential components of clinical care in genetics and what third party payers consider reimbursable services.

   This issue is clearly related to concerns with systems of finance/reimbursement, but represents a more fundamental issue with respect to what constitutes standards of care within genetics, what constitutes a billable unit of time/service for third party payers, and the mechanisms needed to ensure coherence between these two aspects of health care. In the interviews, genetic service providers raised this discrepancy most frequently in discussing the inability to bill directly for genetic counseling services, but also noted the lack of reimbursement for intellectual time, as evidence of a systemic failure to support quality genetic services.

3) An inadequacy in the current supply of genetics professionals, including medical geneticists and genetic counselors.

   Providers felt that the current supply of genetic professionals is not sufficient to meet either current levels of utilization or the expected demand for future services.

4) A lack of effective integration between current systems of non-genetic primary and specialty care and genetics.

   There was a general sense that as genetics becomes increasingly important to the practice of non-genetics primary and specialty care, the lack of effective integration may contribute to inadequate and ineffective patient care.

5) Knowledge, educational, and informational gaps and needs throughout the primary care and non-genetic health workforce.

   Genetic service providers are clearly concerned by what they perceive as a significant lack of appreciation for the growing importance of genetics in health care and deficits in the knowledge and skills of primary care providers and the non-genetics health workforce. Their concerns are motivated in part by what they see as compromises to patient care. A number of providers registered their concern that primary care providers are currently failing to offer standard prenatal screening tests despite clear indicators; are not recognizing risks or referring patients appropriately; are utilizing tests inappropriately or providing inaccurate interpretations and/or clinical recommendations to patients. These issues are of particular concern as many anticipate a growing role for primary care and the greater health workforce, and an increasing need for non-genetic specialists to incorporate genetics into their care giving practices in order to provide patients with the best care possible.
6) Knowledge and informational gaps and needs among the public.

   Genetic service providers expressed concern that a lack of basic genetic literacy among the population may affect utilization and quality of care. There were concerns that technology and the commercial availability of genetic products may outpace the availability of accurate information and public comprehension. These led to additional concerns about consumer protection and the potential for abuses of genetic technology. A few providers also believe that public perceptions, including the conflation of genetics with abortion, were affecting service utilization.

7) Access disparities, including (a) an uneven distribution of genetic service provision, and (b) a lack of linguistic and cultural diversity among genetic service providers.

   While most providers expressed concern with the lack of genetic services in many areas of Illinois, a larger concern for all providers was with ensuring that the benefits of genetics as a component of health care reach those communities that are and have been traditionally underserved by the health care system.

8) Ethical, legal, and social concerns, including issues related to genetic discrimination and concerns that genetics may exacerbate existing or create new health disparities.

   These concerns include insurance and employment discrimination, privacy of genetic information, and direct-to-consumer marketing of genetic tests. Another set of concerns involves the provision of effective counseling to patients, before they undergo genetic testing, regarding the ethical, legal, and social implications of testing.

   As key stakeholders in the genetic services delivery system, genetic service professionals provided insights into the current state of services in Illinois, anticipated future needs in the state, and the key barriers to care. The major themes that emerged were: the need to address access disparities, the need to address the inadequacies in finance/reimbursement, and the need to address knowledge deficits among the public and the primary care and larger health workforce. These also represent the most frequently noted barriers to care, and are highlighted by the role they suggested for the state department of public health, which focused on education, direct service provision, assuring access, and assuring a stable and secure system of finance/reimbursement for genetics. While emphasizing important systemic issues, genetic service providers also stressed the need for the state to continue to support public health’s role in the provision of direct services.
Non-Genetic Health Care Providers

We interviewed 10 non-genetics health care providers: one pediatrician; one board certified family physician; two pediatric neurologists; two pediatric hematologists/oncologists; one pediatric cardiologist; two clinical psychologists; and one midwife. Interviewees represented private practice offices, academic medical centers, and public health clinics. The list for the other health care provider sample was developed through consultation with genetic providers. The types of medical specialists included were determined by knowledge of where genetics currently plays a role in diagnosis and treatment of patients, or where advances in genetics are expected to play a role within the next several years.

This set of interviewees was small and diverse in both their medical specialties and involvement with genetics; therefore, their responses may not be generalizable to others in their specialty area. Although not generalizable, the responses from this diverse group provide important insights into how genetics is being considered and incorporated within the larger medical community.

Interviewees were asked how genetics has impacted their practice in the past five years and how it is expected to impact their practice in the next 5 years. Respondents said they are currently negotiating and expect to contend with an increase in the availability of genetic screening tests, diagnostic tests, and treatment options, as well as more inquiries about and awareness of genetics from patients. The psychologists reported that providers and patients have an increasing awareness of psychological counseling needs for patients both before and after genetic testing. A pediatric hematologist/oncologist noted the importance of advances in pharmacogenomics in oncology. Another specialist expressed a number of concerns about cost and insurance issues, including a fear that the discrepancy between those who can and cannot afford genetic testing is only going to get worse. One of the primary care providers predicted “exponential” growth in genetics and expressed the following concerns: how can primary care providers keep up with and utilize new developments?; will the gap widen between knowledge and application of that knowledge?; and will there be new treatments to go along with the ability to diagnose more conditions?

We asked primary and specialty health care providers about their role in the process of genetic service delivery. Most commonly, interviewees reported that they identify genetic risk and, when appropriate, refer patients for genetic counseling/evaluation. Some specialists provide services to clients with genetic conditions that are referred through APORS or other health care providers/systems, order genetic tests, or may provide genetic counseling when the condition is “straightforward.” One primary care physician described his role in providing follow up care to patients following genetic consultations: “Once a diagnosis is made, [I] serve as a clearinghouse – all the reports come to me…[I] keep people headed in the right direction, making sure they get their follow-up appointments and any supportive care that they might need.” Interviewees also reported educating family members and/or caretakers on a patient’s genetic condition, and writing letters of medical necessity to insurance companies on their patients’ behalf.

Despite indicating a number of concrete ways in which they are currently involved in the provision of genetic services, some respondents expressed uncertainty about their role in the genetic service system. A family practitioner who expressed uncertainty followed that statement by noting, “We are really limited with what insurance allows.” One physician expressed a desire for guidance and leadership:
I think that in order to deliver full-service to patients and families, we really need a big educational push. We need a systematic approach to, and guidelines, as to what kinds of genetic things should we be testing for, how to do that, where should the test be sent, [appropriateness of the test]. I think that there needs to be some person or body to step forward and take the lead in systematizing an approach for the state.

Interviewees were asked to comment on the challenges they perceived in their provision of genetic services. Two respondents mentioned the cost of genetic testing and insurance coverage. Other challenges mentioned related to genetic testing included the amount of physician and staff time required to obtain payment or insurance coverage for genetic tests, difficulties getting test results back in a timely manner, and institutional contracts with laboratories that determine where tests must be analyzed. Education was a major challenge noted by several respondents, including: the difficulty of keeping current on new developments in genetics; the lack of familiarity with symptoms and of practical experience managing certain types of conditions; and the lack of easily acquired genetics knowledge. One provider stated “Genetics is an arcane, mysterious branch of medicine that is often poorly understood by the rest of us. [Geneticists] use a lot of jargon… [Genetics] is an inherently complicated subject…that is challenging not just to describe to laypeople, but it’s challenging even to grasp for medical professionals who are not in the genetics field.”

Other challenges mentioned included the limited number of genetic professionals for face-to-face and phone consultation, and the lack of patient and public education and awareness about the importance of genetics. One specialist noted that while the geneticists and genetic counselor he/she interacts with are responsive, there are just not enough of them. The primary care providers also raised questions concerning the clinical benefit of genetic testing to the patient (i.e., will the information really make a difference in the patient’s life expectancy, quality of life, or disease management).

The most commonly mentioned access barrier, raised by four respondents, was lack of public knowledge and education. The lack of patient “know-how” to maneuver the health care system and problems with patient compliance and follow-through were identified as problems specific to patient education and awareness. Four respondents mentioned lack of insurance coverage/reimbursement as an access barrier, two each mentioned limited supply of genetic providers, need for health care provider education about genetics, and language problems. Other access barriers included transportation, stigma, and a lack of referral from PCPs.

Respondents identified the most needed genetic services and resources, including: (1) a systematic approach to genetic testing, including resolving the current “double standard of care” based on ability to pay; (2) more genetic providers, especially in rural and economically disadvantaged communities; (3) enhanced education, for providers, patients, and the public; and, (4) increased funding for genetic services. Recognizing that there is and may always be a need for more geneticists and genetic counselors in rural areas, and that other providers may have to fill that gap, one specialist stated, “Eventually, primary care physicians, public health nurses, people on the primary care front lines, are going to need to have a pretty good working understanding of genetics and the basics so that they can work with families…I don’t think we have that knowledge base yet or a training program to establish that knowledge base for primary care providers."

Several key themes emerged from these interviews regarding the challenges the state will face in the next five years. These include the knowledge explosion occurring in the field of genetics, the increasing availability of genetic tests, concerns about how genetic testing will be paid for, the limited supply of genetic professionals, and the need for physician education. There was discussion about the importance of and need for a more systemic approach to genetic evaluations other than neonatal screening. This respondent also spoke about the challenge of developing a
genetic service system that is “high quality,” “appropriate,” “non-punitive,” and includes physician education.

There were a number of important comments and recommendations regarding IDPH’s role in the genetic service system, generally regarding service provision, education, and finances/reimbursement, which are discussed below.

• Service Provision
  Three respondents thought that IDPH should be involved with more genetic screening, taking a public health approach to genetic conditions to “do the most for as many people.” In addition, there were suggestions for IDPH to support the placement of genetic counselors in public health departments, facilitate the streamlining of a referral process for providers to make it easier for children to obtain genetic services, establish data bases and store biological DNA to help people keep track of and help other family members, and to have a lab or service to support testing for rare diseases.

• Education
  Seven respondents identified education as a crucial role for IDPH, for the public (e.g., awareness about what genetics is, and about the importance of genetic counseling) and for health care providers (e.g., recognizing disorders, knowing when and to whom to refer patients). One specialist summed up an argument for education as follows:

    I think, for the most part, the public health department…is relatively trusted by the bulk of the population. People view the health department system as something that provides a lot of education and provides a lot of assistance in things that involve the medical/social goodwill of the population. I think using the health department system is an appropriate way to go to try and disseminate education to primary care providers of all sorts as well as the public. I think that the state needs to support that and put the funding in to do that.

    By contrast, another respondent felt that “the state ‘already [does] a fair amount of education and there’s only so much education you can do. You can only educate people who want to be educated. And largely the people who want to be educated are the people who already have the education… I think we need money and the means to get stuff done.’”

• Finances/Reimbursement
  Financial support for genetic counseling training programs, genetic services and/or genetic testing was mentioned several times, including Medicaid coverage of genetic services. One respondent spoke about the inequities in reimbursement for “specialties that are cognitive in nature… Payment is based on procedures, not on the hours of having to do research and discuss with the family. So recognizing intellectual pursuits as just as important as procedures would help me feel like I am valued.”

In summary, this small, diverse group of primary and specialty care providers provided insight into their current involvement with the provision of genetic services in Illinois, the challenges they face, and their thoughts on IDPH’s role in the genetic services system. Genetics has impacted all of their practices and is expected to continue to do so. However, providers reported uncertainty about their place in the genetic service system. They were concerned about insurance and reimbursement issues, education for themselves and the public, and the supply of genetic service providers. Their
recommendations for IDPH’s role in the genetic service system included service provision (i.e., genetic screening), public and health provider education, and financial support.
Local Health Departments

A total of 15 representatives from 14 local health departments were interviewed; half with IDPH genetics grant funding and half without funding. In addition, geographic area was a criterion, based on the seven Illinois regions used by IDPH. Therefore, we selected two health departments from each of the seven IDPH regions, one “funded” and one “unfunded.” At least 13 of the interviewees were nurses.

The respondents from grant-funded health departments shared similarities in their involvement with genetic services, performing the duties associated with receiving the grant: using the IDPH genetic screening tool; following up on clients with positive indicators; referring clients to regional genetics centers; and following up on newborn screening, newborn hearing referrals, and APORS referrals. Some of these held on-site genetic clinics and/or provided outreach and educational services to other health departments in their region. A description of how health departments are currently involved in providing genetic services and referrals is included in Chapter 3: Current Status.

Interviewees were asked about what was and was not working well with the current system of providing genetic services through local health departments. Three funded respondents mentioned the annual genetics conference and three noted their relationship with the regional genetics center as examples of what is working well. One respondent stated “I didn’t really want to deal with genetics when I came here. And I went to the first [genetics] conference and I came back really fired up.”

There were many comments about what was not working well in the current system. Both a funded and unfunded respondent reported client non-compliance (lack of follow-up on referrals, not completing paperwork) as a problem. Unfunded respondents noted the need for more timely and accurate data on APORS, newborn screening and newborn hearing referrals, as well as out-of-county deliveries. Three respondents (funded and unfunded) mentioned that health department staff could use more training in genetics; in addition, one of these respondents noted that funding would be needed to provide this educational training. Other funded respondents indicated that resources are needed to “get the word out” about what genetics is and why it is important. An unfunded health department interviewee stated that her health department does not get reimbursed for “what we do.” In terms of further incorporating genetics into nursing responsibilities there were conflicting statements from unfunded respondents about whether it would be feasible to do so.

Funded and unfunded respondents reported similar challenges with respect to providing genetic services. The most common challenge, reported by five respondents, was transportation and travel problems for clients. Three respondents noted insufficient funding for genetic services as a problem, while time, client non-compliance, and the difficulty in locating clients were each mentioned by two respondents. Other reported challenges included the need to educate and inform nurses about genetics, provide information to the public, find resources for Spanish-speaking clients, obtain a list of genetic resources and providers, help clients improve their recall of family history, and address nurse staffing issues (turnover/recruitment, and the need for more genetic services in the area). One unfunded interviewee discussed the problem of the attitude of local physicians: “We battle with one of our [local] pediatricians really big-time. If we refer [out for genetic services], he gets very angry with us.”

Respondents were asked to think ahead about how genetics will affect their health department’s practices/programs over the next 5-10 years. There was an expectation from many respondents that advances in genetics and greater public education would have a substantial effect on health department programs, whether in new areas or by doing more of what they currently do. For example, one funded respondent had the following perspective, “I think genetics [will] be a huge part of public health because genetics is booming. Everything is genetics-related.” She went on to comment on
the relationship of health education to genetics, specifically regarding cancer, heart disease, and respiratory disease. Other funded respondents voiced concerns about the lack of attention to or discussion of genetics and how it will be applied throughout the health department.

In addition, several respondents, from both funded and unfunded health departments, mentioned that they anticipate people requiring and demanding more genetic services and information. There was concern about staff being able to keep up with the expected increased demand for information, screening, and follow-up services. Other challenges mentioned were the need to address “adult genetics,” difficulty in handling the increased number of babies surviving with more complex conditions, and the need to make people more aware of what is currently offered by the health department in terms of genetic services.

Interviewees were asked their opinion regarding the biggest barriers to access with respect to genetic services and what genetics services or resources were most needed in Illinois. Almost all respondents indicated transportation or travel barriers as needs. As one nurse stated, "I guess it doesn't matter whether you're traveling 50 miles or you're traveling 10 - if you don't have a car, it's a long way to walk." This issue was related to other issues raised by both funded and unfunded health departments about the need for more accessible services and providers in local and rural areas, as well as financial or insurance barriers.

Many funded and unfunded respondents raised the issue of education as both a need and a barrier to access. Two respondents indicated that they felt that lower-income and less educated clients do not view preventive care as a priority. Others noted the following needs: better education about available genetic services for families in and out of the hospital; and genetics education and training for nurses, physicians, public health personnel, and the public about what genetics is, why it is important, and the genetic services available through the health department.

Other access barriers mentioned were language, cultural and ethnic differences, and the complexity of the forms. Two respondents noted that more genetics providers were needed in Illinois, as well as more genetics education pamphlets.

Interviewees were asked their opinion regarding IDPH’s role in the genetic service system in Illinois. Both types of respondents mentioned education and funding as priority roles for IDPH. Three respondents mentioned the role of IDPH in public and professional education, while additional respondents noted the need for IDPH to educate health department nurses, caregivers, and clients. A funded and an unfunded respondent noted the need for downstate educational and training opportunities. There were differing viewpoints on the importance of IDPH’s education role:

"Maybe the priority ought to be to keep on keeping on and not changing a whole lot. Education at this level, if it's anybody's fault, it would be mine. I don't know that the state itself could do anything."

"I feel there is a knowledge deficit [in our health department]. I think that there needs to be intense training."

"I think [IDPH] should be the leaders. I think that they are the ones that need to make everybody aware. I think that they are the ones that need to provide the training and help…I see them as THE leaders of what needs to be done."

Funded respondents also identified the following priorities for IDPH: make funding available to support genetics centers and providers; provide genetic testing for families that cannot afford to pay; assist children already in the system; keep health departments informed of funding and education opportunities; and work to decrease morbidity and mortality rates. Unfunded
respondents also noted the following priorities: provide more grant funding to more rural areas; including transportation assistance; focus on prevention and screening for birth defects; and monitor referrals and hospitals.

The challenges and access barriers identified by interviewees from local health departments appear to reflect the population served by local health departments (generally, poor and uninsured, often rural) as well as broader system issues: transportation problems; client non-compliance; lack of client education/awareness about genetics and genetic services; and financial/insurance constraints. Local health department interviewees also reported a need for more staff training in and awareness of genetics. The most frequently mentioned roles for IDPH were education (public, caregivers, health department staff) and funding (genetic testing, genetic services, transportation assistance).
State Agency Personnel

Twenty-one individuals from state agencies (IDPH, DHS, DSCC) were interviewed for this needs assessment. Respondents represented a variety of chronic disease, maternal and child health, and other health programs as well as the genetics and newborn screening program. Their experience, education, current position and responsibilities, and knowledge about genetics varied considerably. Many of the interview responses were specific to the program areas in which the respondent worked. This analysis is meant to illuminate the thoughts and concerns of the state agency personnel we interviewed without the detail that would allow readers to identify the responses of particular interviewees.

Regarding their genetics knowledge and training, more than half of respondents indicated that they had very little experience and/or education in genetics. Even among those who answered that their knowledge was average or above average, many reported that this experience was from on-the-job training.

Outside of the genetics and newborn screening program, genetics was a minimal or nonexistent part of most program areas. However, chronic disease program staff often noted that they were part of the Genomics and Chronic Disease Prevention Team. There were also some surveillance activities, such as APORS, that included genetic disorders. Another respondent noted that they do not address genetics because of lack of personnel resources.

Interviewees were asked how they thought genetics information might now or in the near future benefit their program and clients and what opportunities they saw for integration of genetics into their program area. The responses ranged from feeling that integration is unlikely to “I see a lot of opportunity… I think we could do more and do better.” Many responses addressed the need for more education, awareness and information for their staff and/or clients in order to better integrate genetics with their program area.

Respondents in various ways indicated that organizational structures and funding streams were barriers to integrating genetics into their program areas. Regarding organizational structure, one respondent said “we are all so busy doing what we have to do, it’s hard to look at what we should be doing” and referred to the structure of the public health department in terms of “silos.” We also heard from two respondents about the influence of funding agencies: “We have to respond to our funding agent… in general, they encourage integration with other programs but they do not specify that genetics has to be involved.”

Several state agency staff reported the lack of “time, staff and money.” In addition, another reported barrier was the lack of genetics education - of program staff, program administrators, agency administrators, the public, and clients and parents. One interviewee said that state agency personnel first need to educate themselves about the importance of genetics, and then a public relations initiative is needed. Another noted the need for the professionals involved to make genetics a priority and develop action plans for integrating genetics into health care in a way that will serve the neediest patients.” Other barriers included: public attitudes and fear of how insurance companies may use information; a need for staff training and a mechanism to pay for that training; and systemic constraints.

There were a variety of responses from state agency staff regarding the major genetics or genetics related issues that public health will need to address in the next 5 years. The most commonly reported issue was that of genetics education. Four respondents each noted the need for educating primary care providers and/or non-genetic health care providers, four respondents also indicated the need for public education, and one respondent mentioned the need to educate public health nurses about genetics. Other educational issues raised were “raising awareness of the relationship between genetics and the risk for chronic disease,” and incorporating that into health
promotion educational messages, as well as noting the importance of following up on family history and of relating it to chronic disease. Funding issues were also identified by two respondents, with one stating “public health is always pennies on the dollar in terms of health care.”

Three state agency interviewees also raised workforce concerns, specifically that there is a shortage of genetic counselors, and that there are not enough genetic providers to deal with expected increases in the volume of people screened and cases identified (e.g., from universal first trimester prenatal screening or expanded newborn screening). Another respondent questioned the trend of expanded screening in cases where there are no interventions, stating that even for the conditions for which there might be some interventions, “will we have the resources to provide [the needed services]?”

Specific health areas that may need to be addressed in the next five years were also mentioned, including obesity, cardiovascular health, hypertension control, and chronic disease in general. The following issues were raised by one respondent each: concerns about insurance discrimination; invariable access; prenatal exposures; gene-environment interactions; early identification of genetic disorders in children; and tight school budgets but more mainstreamed children with genetic disorders who require nursing intervention. One respondent noted that it will be a challenge to "balanc[e] the people who want to know everything and want every possible test with what is cost-effective."

State agency interviewees had a variety of opinions about what genetics services or resources are most needed in Illinois. The most commonly mentioned need identified as mentioned above, was for more genetic providers (medical geneticists, genetic counselors, medical geneticists, genetic counselors) and for more providers in underserved areas (e.g., southern Illinois) to ease access and transportation problems, especially if more screening tests are added (e.g. cystic fibrosis) or if/when genetic screening and counseling for chronic diseases becomes more common. One respondent stated "Generally speaking, the resources are out there. It's a matter of identifying them and putting them into some sort of a database that you could share that information with people." Another respondent also identified the need for the state to develop a “resource center” for providers.

Other responses from state agency interviewees about resource needs included: financial issues (lack of insurance, limited access for poor and uninsured individuals); education of health care professionals; adequate funding and staffing; public education to “clear up myths;” understanding of risks and risk differences by ethnicity; social and psychological support; and bridging public and private services. One interviewee noted that working with the University of Illinois at Chicago (UIC) School of Public Health could help “get the word out.”

The most commonly mentioned access barrier, with respect to genetic services in Illinois, was financial access/lack of insurance coverage (mentioned by seven state agency respondents). Lack of knowledge and awareness were also mentioned by numerous respondents in different contexts, such as a general lack of knowledge, lack of knowledge about where to go for genetic services, and the need to make the appropriate people aware of available services. Similar to the previous two sections, the lack of genetic providers outside of Chicago was stated as an access barrier by two respondents, with another state agency respondent noting not enough genetic providers in general.

Other access barriers mentioned include: client failure to follow up; lack of transportation and far distance to genetic services; poor access for poor and uninsured; fear about how genetic information might “come back to haunt someone;” lack of funding to pay for diagnostic work-ups; lack of routine genetic testing as part of most disease investigations; and the unavailability of data.

State agency interviewees were asked their opinion regarding the role of IDPH with respect to the genetic service system in Illinois. One respondent summed up the overarching role as follows: “…coordinating, integrating and using it's authority as the state health agency to assure that...
the entire population has access to genetic services in the state," with two other respondents voicing similar views about the need to ensure adequate and accessible services. Four respondents stated that IDPH should retain and possibly expand existing programs (e.g., regional genetic center and local health department grant programs). The need for IDPH to work with local health department staff and for all local health departments to use the genetic screening tool and offer genetic referrals was also mentioned.

A consistent theme from state agency interviewees regarding the role of IDPH was educating and providing information to physicians and other health care providers (six respondents), the public (five respondents), in general (four respondents), and to other entities that serve families (one respondent). One interviewee stated, “Education is [IDPH’s] priority.” A few respondents mentioned data and surveillance roles, such as the need to better integrate surveillance data (e.g., with APORS), examine and disseminate data, gather information; assure that hospitals report data, and assure the privacy of health data. Other responses to this question included: subsidize transportation; provide incentives for genetic counselors to work in underserved areas; and integrate genetics into the perinatal care system.

It should be remembered that although this group of interviewees often spoke directly about their program areas, much of this detail was by necessity excluded from the report to ensure confidentiality. The broader outlines that are reported here, however, offer insight into the ways in which genetics is, or is not, currently considered and integrated by different state programs.

Excluding those participants from the state’s genetics program, most interviewees from state agency programs reported having only minimal genetics knowledge and training and reported that their program areas included genetics only in limited ways. Interviewees discussed the need for program and agency staff to have greater awareness and education about genetics. They noted that, in addition to staff education, organizational structure and funding streams were barriers to integrating genetics into their programs. Overall, state agency staff indicated that IDPH’s role should be to coordinate and ensure access to genetic services, educate and provide information to health care providers and the public about genetics, and to assume various data and surveillance functions.
Consumer/Advocacy Organization Representatives

Interviewees included eight individuals representing seven advocacy groups in Illinois. The advocacy groups were varied, with some focusing on genetic disorders and others representing chronic disease or more general health advocacy. There were two types of respondents among the advocacy groups. One was the “organizational leader/parent” who, as a parent of a child with a genetic condition or special health care need, became involved for personal reasons (three respondents). The other was a “professional” within the organization. We should note that this is a small sample from which to draw generalizations or conclusions. We extended invitations to 29 consumer/advocacy organizations but only seven agreed to have a representative participate. Some of the reasons given for declining were: that “genetics is not even on their radar screen;” not knowing who in their organization would be appropriate to participate; and that their national organization had not yet set policies regarding genetics.

When asked to describe their knowledge and experience in the field of genetics and genomics, responses ranged from “limited but learning” to slightly above average and above average. Experience in genetics/genomics included none, preparing and mailing out information to families, and/or referring individuals who have a genetic condition to genetic professionals.

Interviewees were asked to describe their organization’s involvement with genetic services in Illinois. Participants described their agency’s involvement with IDPH and other state agencies as ranging from none at all, to receiving some funding for various projects, to working more extensively with IDPH on task forces, state committees, and educational and policy making related to genetics. Participants responding from organizations advocating for specific genetic conditions tended to describe more active involvement with genetic professionals, including establishing and maintaining relationships with genetic counselors and geneticists in Illinois. One organization provides funding for projects related to genetics. Other agencies provide resources about where to go for genetic services; offer educational assistance about genetic issues; and talk with families about genetics. One agency representative indicated that they have only “peripheral” involvement with genetic services.

When asked how the agency communicated with its constituency about genetic issues, responses varied from minimal and informal communication to more extensive, direct, and formal patterns of communication. One respondent described how they distribute literature, including fact sheets about genetic issues, through health departments and clinics throughout the state, and include genetics-related information in newsletters and annual reports. One respondent noted that her agency refers people to a national hotline and answers questions “to the best of our abilities,” but reported that they do not get many questions. Two advocacy group representatives noted that they try to get access to patients and families through practitioners, and try to get the services out to people, but they need “some sort of consistent link to services.” Two representatives described limited communication with their constituents about genetic issues.

Key areas of concern for consumers in using genetic services were explored with the interviewees. Among the concerns raised was the issue of confidentiality of personal genetic information, and the potential for employment and health insurance discrimination. One interviewee noted her perspective that Illinois non-discrimination legislation lacks adequate enforcement provisions. Another main concern was obtaining accurate information and knowing where to go once a genetic risk is identified or a diagnosis is made. Other concerns mentioned were understanding what genetic services are, as access to care, and health coverage.

Interviewees identified several strategies to address these concerns. These included:

- **Education**
  - educating clinicians so they can respond when patients have genetic questions;
  - educating the public – informing them of the availability of support services;
educating health advocates so that each agency will have an identified expert who is knowledgeable about resources and how to answer genetic-related questions; and,
• providing clear messages and information about what genetic services are and how to access them.

• **Policy strategies**
  • addressing people’s fears about having genetic testing; and
  • addressing concerns about insurance discrimination.

• **Legislation**
  • raising the allowable parent income level for Title 21 Family Care and allowing parents to continue coverage when a child has special health care needs or is on Supplemental Security Income (SSI).

Interviewees were asked what they thought would be the **major genetics or genetics related issues** that public health will need to address in the next five years. The responses were similar to the strategies stated above: the need for educating the public and the health care workforce - it was noted that it must be difficult for genetic professionals to keep current with all the new advances and educate the public too; an increasing need for support services both for individuals and families with genetic conditions and for people who work in advocacy organizations; and changes with health insurance coverage to ensure that there is equality of care. One respondent mentioned the importance of reimbursement for genetic services, and the need for more programs for genetic counselors and/or having other clinicians cross-trained in genetic counseling.

Genetics services and **resources identified by interviewees as in greatest need in Illinois** included:

• More resources – staff and technology - for expanded newborn screening.
• More resources, workshops, and conferences for families who have a child with a genetic condition so they “can connect” with one another.
• A “one-stop” resource for genetic information on a local level.
• Mandates against insurance discrimination, and for insurance to cover needed services.
• Access to genetic services, especially for those from other cultures and who speak another language (address language barriers and cultural competency issues)
• More education for the public about what genetics is and what it has to do with their daily health.

A number of **barriers to access to genetic services** in Illinois were identified by interviewees:

• Cost and adequacy/lack of health insurance coverage.
• Fear of insurance and employment discrimination on the part of the public.
• Reimbursement of genetic services
• Lack of proximity of genetic services – distance as a barrier.
• Physicians and hospital staff: 1) Lack of culturally competent care ; and 2) lack of willingness/consistency in offering genetic support resources to families.

Interviewees identified a number of activities and roles that IDPH could assume with respect to the genetic service system in Illinois. Educating providers about genetics, communicating with patients about genetics, and providing parents with information about support organizations were indicated as very important by two interviewees. One respondent said they would like to see more support –more funding, resources, and a better data system/technology – from IDPH leadership for the IDPH genetics program. Another respondent said that IDPH refers requests for
prevalence/incidence data to her organization when the state should really be tracking this information. Expanding newborn screening and being an informational resource to support organizations were also suggested. One interviewee suggested that IDPH survey hospitals to identify the extent and availability of hospital-based genetic services and, if a system is not in place, ensure that one is created so that patients can have access to genetic services.

In this study sample, the focus of persons in the role of organizational leader or parent tended toward personal issues and the need for support services; how well health care providers communicate with families about genetic issues; and concerns about the accuracy of the information provided by health care professionals. Respondents in the role of advocacy professional tended to focus on education and training for themselves and for health professionals, as well as on the need for policies to protect the privacy of genetic information. Continuing education for clinicians and health care providers was already a service provided by two of the advocacy groups.
Chapter 7: Public Attitudes and Perceptions of Ethical, Legal and Social Issues (ELSI) in Genetics

We did not directly survey the public as part of this needs assessment process. The attitudes and perspectives of both the general public and specific populations affected by genetic conditions have been well studied by independent researchers and states alike. However, even as majority positions emerge among members of these groups, strong minority positions remain. Infused with people's enthusiasm for the prevention and health promotion benefits of medical genetics is an undercurrent of moral and ethical concern about how new genetic technologies should be applied in medicine, insurance, employment, and government regulation.

Key Findings

Based on our review of numerous studies (see Table 7.1 at the end of this chapter), we draw several conclusions about the public’s perceptions of, and attitudes towards, genetic technologies in healthcare markets. While this literature review is not exhaustive, and our findings may therefore not be representative of the entire U.S. population, it does provide a useful starting point for crafting representative genetics policy. The following is a summary of the key findings, which will be expanded on below.

- People are increasingly aware of the role of genetics in medicine, but do not completely grasp its complexities.
- Emergent majorities believe the application of genetic technologies is a personal decision, but sizable minorities still support complete genetic technology bans.
- Men, political Democrats, Hispanics, and those without a religious affiliation are most likely to support genetics. Income and education do not play a role in willingness to undergo testing, and race seems to play a more complex role.
- At least some of the ambivalence towards genetic technology among the public can be traced to fears of discrimination in employment or insurability.
- Americans are concerned about genetic discrimination and would like to see these issues addressed in discrimination laws that apply to insurability and employment.
- The public believes that all segments of the population should have equal access to health care that includes medical genetic services. Central state offices should aid the public in locating genetics resources and improving knowledge.
- Individuals with genetic conditions and/or their caretakers should be consulted in policy decisions that will affect them. These populations may have strong opinions about how to improve service delivery.

The North Suburban Library System Study

To date, the most extensive study of Illinois residents' knowledge of and attitudes towards genetics has been conducted by the North Suburban Library System (NSLS)\(^\text{167}\). Comprised of libraries in the north and northwest suburbs of Chicago, the NSLS conducted twelve focus groups with 131 individuals in 2001. Participants discussed the impact of genetic technology both before and after viewing a short videotape about the Human Genome Project. They found that participants:

• were mostly aware of the Human Genome Project and/or human genetics, and strongly associated these with advances in medicine;
• were concerned about privacy and discrimination issues;
• were inquisitive about the economics of genetics, such as issues of ownership of the genome and regulation of for-profit biotechnology businesses; and
• believed the impact of the human genome project could be very beneficial, but were also generally uneasy about the potential effects of “tampering with nature” and were concerned about people’s emotional well-being following testing.

The NSLS study serves both as a useful abstract to summarize public attitudes in general, and as an important start for understanding Illinoisans’ attitudes towards genetic health policy.

Public Awareness and Knowledge

There is some evidence that the completion of the Human Genome Project in 2003 brought heightened awareness of the medical implications of genetic technology to the general public. Comparisons of historical data indicate that the public knows more about genetics than it did a decade ago. For example, during the early 1990s, the state of Washington reported that only 6% of all respondents to a telephone survey of its residents had even heard of the Human Genome Project. However, a recent national survey indicated that at least 80% of Americans had at least heard of cloning, in vitro fertilization, genetic testing, and prenatal testing.  

Increasing public awareness of genetics does not necessarily translate into an understanding of the complexities in this area or of the medical subspecialty field. Several states have indicated their residents are uncertain about the organization of their genetic services delivery systems. Other studies have indicated that the public is more likely to be aware of only well publicized genetic discoveries, such as those associated with breast and colon cancer. Very few people have more than a simple understanding of complex environment/gene interactions and other less well publicized aspects of the science. However, one notable study did find that several focus groups of Georgia residents understood that “a gene for heart disease” was most likely to mean that genetics worked in combination with environmental factors in triggering heart disease.


172 Genetics & Public Policy Center.


174 Genetics & Public Policy Center.


Public Attitudes

Many studies mention that the general public is enthusiastic about the life-saving potential of genetic technologies. Likewise, several studies have shown U.S. residents are willing to undergo genetic testing for a range of prenatal, newborn, and adult-onset genetic conditions. However, sizable segments of the population still remain that do not support current technologies or have serious reservations about their use. Twenty percent of respondents to one national survey felt that pre-implantation genetic diagnosis should be completely banned, and 11% felt the same way about prenatal testing. However, such reservations may be more limited to prenatal genetic practice than to non-reproductive adult genetic services, and may vary by racial or ethnic group for at least some adult-onset conditions.

The Demographics of Support

Studies have shown that men, political Democrats, and people without religious affiliations are most likely to support genetic technology and its application. Additionally, it appears Hispanics/Latinos are more likely than Caucasians and African Americans to support genetic testing, although African American perspectives appear to be more complex and subtle than is evident in some forced-choice surveys. Education and income seem to have an effect only on a person's awareness and knowledge of genetics, but may still interact in complex ways with willingness to undergo testing.

178 Genetics & Public Policy Center.
190 Genetics & Public Policy Center, 2004.
192 Ibid.
193 Mogilner et al., 1998.
195 Morris et al., 2003.
196 Hicken et al., 2004.
197 Bassett et al., 2004.
Fear of Discrimination

Several studies have found that large majorities of Americans are concerned about genetic discrimination and want anti-discrimination laws to apply to insurance and employment. One study reported that 40% of primary care patients feel genetic testing is not a good idea because of fears of insurance discrimination. However, another study shows that though fear of discrimination is widespread, patients will not let it interfere with their desire to have testing performed.

The evidence is unsettled concerning racial/variation in these fears: It is unclear whether these fears and concerns are more pronounced among ethnic minorities. Although at least one prominent study has reported that ethnic minorities appear especially concerned that they will be targeted for discrimination, other studies have shown that at minimum, racial minorities overall show more variation in concerns about discrimination than was originally reported. For example, one large survey of primary care patients in the U.S. found African Americans are least likely and Hispanics are most likely to be concerned about genetic discrimination in insurability. Another study demonstrated that the fear that genetics will be used to further social inequality is widespread among several ethnic groups.

Fears of discrimination pose a challenge for health care policy makers since they affect use of available genetic services. People who are concerned about discrimination are more likely to pay for testing out of pocket than utilize insurance, refuse testing, or use an alias when requesting testing. These individuals may also request that results be excluded from medical records.

The Service Delivery System

The public is concerned that health care financing policies will create, or is currently creating, inequality of access to emerging genetic services along racial and/or socioeconomic lines. A full 100% of respondents to a survey conducted by the state of Michigan felt that the state should use all available resources to ensure equal access to genetics. This finding has been replicated on a national level, albeit to a smaller extent.

Additionally, the residents of several states have expressed interest in ensuring that consumers, health care providers, and policy makers are educated about emerging genetic science.

---

204 Schulz et al., 2000.
205 Hall et al., 2005.
206 Singer et al., 2004.
210 Schulz et al., 2000.
Consumers would like central state offices to provide information and resources about genetic health care to affected individuals. The public does prefer at least some regulation of genetic technology. Although majorities of the public do not want government interference in private reproductive and healthcare decisions, bipartisan majorities have been found to support governmental efforts to regulate the safety of genetic testing.

Individuals with Genetic Conditions and Their Caretakers

The array of opinions on genetic testing among individuals with genetic conditions is diverse. Several studies have found that populations with more serious genetic disorders, such as dwarfism or mental retardation, are more likely to support policies that aid quality of life, such as screening for at-risk parents, rather than population-based screening services. Similarly, one study showed that as many as one-half of deaf college students did not agree with population-based genetic testing on infants for deafness genes.

State studies have shown that families of children diagnosed with genetic conditions spend enormous amounts of money, time, and emotional and intellectual energy in caring for their children and understanding their child’s specific diagnosis. These families appreciate any additional state resources to help them care for their children, especially aid in locating services and information on genetic conditions. Families want to advocate for their loved ones by choosing between various health services and social/financial supports, but may need additional state assistance in understanding what options and services are available to them.

Conclusion

Surveying the public directly was beyond the scope of this needs assessment process. Our method for gauging attitudes and perspectives about ethical, legal, and social issues in genetics was to study secondary sources. Many studies are referenced above. The planning process will elicit input from Illinois residents on these vital issues.

---

211 Michigan Department of Community Health, 2002.
213 Texas Department of Health, 2002. The development of the Texas state genetics plan and a plan for integrated data infrastructure for genetic services. Available at http://genes-r-us.uthscsa.edu/resources/genetics/texas_geneticsplan.pdf
214 Michigan Department of Community Health, 2002.
216 Campell & Ross, 2005.
217 Skinner et al., 2003.
220 Texas Department of Health, 2002. The development of the Texas state genetics plan and a plan for integrated data.
222 Ibid.
<table>
<thead>
<tr>
<th>Title</th>
<th>Authors</th>
<th>Date</th>
<th>Methods</th>
<th>Key Findings</th>
</tr>
</thead>
</table>
• Only 6% had heard of the Human Genome Project; a large majority had heard “relatively little” or less about population-based genetic screening.  
• After hearing a brief description of the HGP, a large majority indicated it would be helpful in general to people. |
| 1999/2000 Focus Group Content Report: Communities of Color & Genetics Policy Project | Schulz et al.                                                           | 2000    | Nine focus groups conducted with African American and Latino participants from across a range of income levels; focus group participants from Tuskegee, AL and several cities in Michigan. | • Widespread optimism about the benefits of genetic technology is tempered by concern about people's ability to understand genetic advances.  
• Skepticism about the motives of the scientific community in promoting advances, and over who will control genetic information. |
| Democratizing Human Genome Project Information: A Model Program for Education, Information and Debate in Public Libraries | North Suburban Library System (Metro Chicago, IL)                      | 2001    | 12 focus groups held in 2001 in north suburban Chicago area; 131 participants recruited by librarians | Respondents were:  
• Mostly aware of the Human Genome Project and/or human genetics, and strongly associated these with advances in medicine.  
• Concerned about privacy and discrimination issues.  
• Inquisitive about the economics of genetics, such as issues of ownership of the genome and regulation of for-profit biotechnology businesses.  
• Believed the impact of the Human Genome Project could be very beneficial, but were also generally uneasy about the potential effects of “tampering with nature” and were concerned about people's emotional well-being following testing. |
| Alaska State Genetics Plan                                            | Alaska Department of Health and Social Services; [http://hss.state.ak.us/dhcs/PDF/FinalDraft07-28-03.pdf](http://hss.state.ak.us/dhcs/PDF/FinalDraft07-28-03.pdf) | 2002    | Surveys mailed to patients in state supported genetics clinics and handed out at health fairs to members of general public; low response rate to surveys handed out at health fairs. | • About 75% of the general public was at least somewhat concerned about discrimination.  
• Only 15% would know how to access genetics services if necessary.  
• Majorities would elect to have genetic testing performed, and large majorities would use information from testing to make lifestyle changes. |
<p>| Perceptions of Genetics Research as Harmful to Society: Differences Among Samples of African-Americans and European-Americans in Kentucky | Furr                                                                   | 2002    | Random telephone survey of 852 African Americans and European Americans in Kentucky | • African Americans more likely to think that genetics is harmful to society as a whole; demographic characteristics and self-described knowledge of genetics had no effect on African American attitudes. |</p>
<table>
<thead>
<tr>
<th>Title</th>
<th>Authors</th>
<th>Date</th>
<th>Methods</th>
<th>Key Findings</th>
</tr>
</thead>
<tbody>
<tr>
<td>European - Americans</td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Title</td>
<td>Authors</td>
<td>Date</td>
<td>Methods</td>
<td>Key Findings</td>
</tr>
<tr>
<td>----------------------------------------------------------------------</td>
<td>------------------------</td>
<td>--------</td>
<td>-------------------------------------------------------------------------</td>
<td>-------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------</td>
</tr>
<tr>
<td>Psychosocial Predictors of BRCA Counseling and Testing Decisions Among Urban African-American Women</td>
<td>Thompson et al.</td>
<td>2002</td>
<td>Mail survey of 76 African American women with positive family history for breast cancer. Free BRCA counseling and testing were offered to all participants.</td>
<td>- Participants who did not participate in counseling anticipated more negative emotional consequences to testing and more concern about stigmatization.</td>
</tr>
<tr>
<td>What Does ’A Gene for Heart Disease’ Mean? A Focus Group Study of Public Understandings of Genetic Risk Factors</td>
<td>Bates et al.</td>
<td>2003</td>
<td>13 focus groups; 108 participants nominated by three community boards in Georgia to represent breadth of community perspectives</td>
<td>- Participants’ interpretation of 'gene for heart disease' did not coalesce around a single interpretation and did not support experts' fears of widespread genetic fatalism.</td>
</tr>
</tbody>
</table>
| Differences between African Americans and Whites in Their Attitudes toward Genetic Testing for Alzheimer’s Disease | Hipps et al.           | 2003   | Convenience sample of 452 respondents from metropolitan Atlanta         | - Although both racial groups showed general interest in testing for Alzheimer’s disease, African Americans showed somewhat less interest.  
- African Americans foresaw fewer negative consequences from a positive test result.  
- African Americans saw fewer overall reasons to get testing performed.                                                                                                                                         |
| Public Knowledge Regarding the Role of Genetic Susceptibility to Environmentally Induced Health Conditions | Morris et al.          | 2003   | 2353 respondents to national survey which asked participants if a person’s genes can make him/her more likely to: (a) have side effects from drugs; (b) develop diseases caused by cigarette smoking; (c) get infections such as the flu; and (d) develop illness from environmental exposures such as pesticides | - 9% believed genetics plays a role in all four conditions; 32% believed genetics plays no role at all.  
- Respondents did not understand that genetics may play a role in illnesses with complex gene/environment etiologies.  
- Increasing awareness of the role of genetic factors in these conditions will be necessary to translate gene discovery into effective personal and public health actions.                                                                 |
| Screening for Fragile X Syndrome: Parent Attitudes and Perspectives  | Skinner et al.         | 2003   | National survey of 442 parents of children with Fragile X.               | - Only 1% of respondents felt that genetic testing should not be offered to parents at any time.  
- About 2/3 agree that testing should be offered during the prenatal period to parents and fetus.  
- Parents more likely to see more advantages than disadvantages to widespread testing and early diagnosis of Fragile X.                                                                                           |
<p>| Perceptions of Genetic Discrimination among At-Risk Relatives of      | Apse et al.            | 2004   | 470 person survey of patients in a hereditary colon cancer clinic located in Baltimore, MD. | - About half of respondents were highly concerned about genetic discrimination; those who were highly concerned were more likely to pay out of pocket for genetic testing.                                                                 |</p>
<table>
<thead>
<tr>
<th>Title</th>
<th>Authors</th>
<th>Date</th>
<th>Methods</th>
<th>Key Findings</th>
</tr>
</thead>
<tbody>
<tr>
<td>Colorectal Cancer Patients.</td>
<td></td>
<td></td>
<td></td>
<td>- Awareness and understanding of laws about genetic discrimination were low.</td>
</tr>
</tbody>
</table>
| The Role of Accuracy in Predicting Acceptance of Genetic Susceptibility Testing for Alzheimer’s Disease | Bassett et al.                                                         | 2004     | 518 respondent survey of offspring of patients enrolled in a genetic linkage study for families with multiple members affected by Alzheimer’s disease.* | - 20% of respondents would refuse a test with zero error rates and 60% would refuse a test with high error rates in both directions.   
- Male gender, low education, and a high perceived lifetime risk of Alzheimer’s were associated with acceptance of high error rates. |
| Reproductive Genetic Testing: What America Thinks (2004)              | Genetics & Public Policy Center at Johns Hopkins University              | 2004     | Pinnacle summary of multiple studies: 21 focus groups with 181 members of general public nationwide; 62 in depth interviews; 2 surveys with over 6000 respondents; 21 town hall forums | Majorities of survey respondents felt it: 
- was appropriate to use reproductive genetic testing to avoid life-threatening diseases or to test embryos for tissue that matches a sick sibling. 
- would be inappropriate to use (hypothetical) genetic testing to select for intelligence. 
- Sizable minorities disagreed with the majority opinion on both these issues. 
- was appropriate to use preimplantation genetic diagnosis to select for a baby's sex. 
- Americans considered a person’s motives, means, alternatives, and long-term consequences when considering the moral appropriateness of using genetic technology. 
- Americans were fearful of the consequences of genetic testing – a world where parents try to prevent the birth of imperfect children and where only the poor have diseases. 
- Americans expected oversight to ensure the safety, accuracy, and quality of genetic testing, but didn’t want government regulations to interfere with private reproductive and healthcare decisions. |
- Over 60% of participants wanted government regulation of access to genetic information. 
- At least 70% of respondents agreed that anti-genetic discrimination laws should apply to medical, disability, and long term insurance as well as to employment. |
<table>
<thead>
<tr>
<th>Title</th>
<th>Authors</th>
<th>Date</th>
<th>Methods</th>
<th>Key Findings</th>
</tr>
</thead>
</table>
| Attitudes about and Psychosocial Outcomes of HFE Genotyping for Hemochromatos is | Hicken et al.     | 2004  | Structured interviews conducted in Birmingham, AL with (a) 87 patients who had undergone HFE genotyping and (b) 40 controls who had not. | • Most participants believed that genetic testing is beneficial.  
  • Controls expected to feel more anxiety than was actually felt by patients. |
| Racial and Ethnic Variations in Knowledge and Attitudes about Genetic Testing | Singer et al.     | 2004  | National telephone survey of 1232 households.                            | • Latinos were more likely to express a preference for genetic testing than non-Hispanic-, African-, and European-Americans, but may hold other beliefs that override these preferences.  
  • African Americans and Latinos were less knowledgeable about genetics than non-Hispanics whites.  
  • Fear that genetics can be used to spread/create social inequality was not confined to racial/ethnic minorities. |
| Attitudes of Deaf Individuals towards Genetic Testing                | Taneja et al.     | 2004  | 64 respondents to survey of deaf college students in Washington, DC.     | • Approximately equal numbers felt that genetic testing for hearing loss should/should not be tested for at birth.  
  • A majority was at least “somewhat interested” in using genetics to select for a mate. |
| Parental Attitudes and Beliefs Regarding the Genetic Testing of Children | Campbell & Ross   | 2005  | A total of 12 focus groups lasting 2 hours; focus group participants were Chicago residents in the University of Chicago Hospitals catchment area. | • Most parents wanted access to genetic tests for their children as they become available and were concerned that all parents be able to access testing equally.  
  • Parents also showed concern about genetic discrimination, but did not let it interfere with desire for testing.  
  • Most parents viewed themselves as the final arbiter of whether the child should be tested, including for adult onset conditions.  
  • Groups consisting of only Black respondents were more willing to allow breaches of doctor-patient confidentiality in informing relatives of genetic risks. |
| Community Involvement in Developing Policies for Genetic Testing: Assessing the Interests and Experiences of Individuals Affected by Genetic Conditions | Gollust et al.    | 2005  | Survey of 189 individuals with achondroplasia and 136 of their first degree relatives.* | • Large majority (90%) supported prenatal testing for at risk parents; only a minority supported population-based genetic screening.  
  • Individuals who judged the condition as being more severe were more likely to support population screening.  
  • Individuals with lower quality of life were more likely to support prenatal testing by affected individuals.  
  • Respondents supported genetic policy that aids their quality of life, rather than policies that implement population-based screening. |
<table>
<thead>
<tr>
<th>Title</th>
<th>Authors</th>
<th>Date</th>
<th>Methods</th>
<th>Key Findings</th>
</tr>
</thead>
</table>
| Concerns in a Primary Care Population about Genetic Discrimination by Insurers | Hall et al. | 2005 | Measures agreement among 86,000+ primary care patient undergoing screening for hemochromatosis with statement “Genetic testing is not a good idea because you might have trouble getting or keeping your insurance.” Patients recruited from five states plus one Canadian province. | - 40% overall agreed with the statement.  
- African Americans and Asians were least likely to express concern about insurance discrimination; Hispanics were most likely to express concern.  
- Lack of high school diploma and college degree were associated with greater concern. |
| Knowledge, Attitudes, and Utilization of BRCA1/2 Testing Among Women with Early-Onset Breast Cancer | Peters et al. | 2005 | 248-Respondent survey of women diagnosed with breast cancer before age 50.* | - Women who had undergone BRCA testing were younger and more likely to be college educated.  
- Knowledge of BRCA testing was high but uptake was low: lack of information about how testing might influence healthcare decisions, fear of the genetic testing procedure and its costs as well as false positive results, were all associated with low uptake of testing. |

* Study methodologies unclear about exact demographics of sample; presumably nation-wide.
Chapter 8: State Laws and Legislation

Illinois has enacted several pieces of legislation related to genetics health policy over the last several years, including the Genetic Information Privacy Act of 1997 (GIPA) that broadly protects the confidentiality of genetic information, and the recent licensing of genetic counselors. In addition to GIPA, the following is a review of major state genetics legislation activity since the 91st General Assembly session, beginning January 1, 1999. A summary showing all legislation, major and minor, can be found in Table 8.1 at the end of this chapter.

Genetic Information Privacy Act & Genetic Privacy Information

The Illinois State Legislature passed the Genetic Information Privacy Act in 1997 (Public Act (PA) 90-0025). This bill guarantees the confidentiality of genetic testing and information derived from genetic testing. The results of genetic testing may only be released to the patient tested and/or the patient's authorized representative. No one may disclose, or be forced to disclose, for any reason either the identity of any person on whom a genetic test has been performed or the results of a genetic test that would identify an individual.

Furthermore, PA 90-0025 prohibits health insurance companies from seeking information derived from genetic testing in order to make policy decisions about health or accident coverage. Individuals can submit their own genetic test results to insurance companies for consideration; however, that information may be considered for insurance policy decisions only if the result is favorable to the individual. More recent legislation has strengthened the GIPA. PA 92-0430, enacted in 2001, strengthens the prohibitions against insurers using genetic testing information. This bill inserts a clause into GIPA that “regardless of the source” of the information, insurers cannot use genetic information for non-therapeutic purposes.

A further strengthening of PA 90-0025 was attempted during the 93rd legislative session. House Bill 4201 mandated that the waivers of health history information used by insurance companies should include a statement explicitly stating that genetic information is not included in the waiver. However, this bill was delayed in committee and has not yet been resubmitted during the current 94th session.

Employer restrictions are more lenient than the restrictions that have been imposed on insurers, though they are specifically and explicitly required to handle genetic information as confidential. No employer can compel individuals to release genetic information nor may employers release that information to other parties, if they have access to it.

Several exceptions to the statute exist. Law enforcement agencies are granted the authority to receive identifying information from genetic testing without consent; this information is admissible in criminal court cases as well. Medical professionals performing organ donor transactions may see the results of genetic testing. State and local health agencies can release identifying genetic information only in very specifically defined situations, such as when proper individual consent has been obtained. Finally, any genetic information that indicates an individual has a disease at the time of testing, whether or not the individual is currently symptomatic, is not subject to the confidentiality requirements of the legislation.

PA 91-0549, enacted in August 1999, aligned the state insurance code with the GIPA. Insurers are prohibited from considering genetic information to be a health condition “in the absence of a diagnosis of the condition related to that genetic information.”

It is important to note that while some genetic information is theoretically exempt from confidentiality, the exemption would in practice have very limited effect. There are very few circumstances in which a genetic test would be used for diagnostic purposes. For example, all genetic testing for hereditary cancer susceptibility syndromes would be confidential. Possible
exceptions include diagnostic genetic testing for Huntington’s disease and other neurodegenerative disorders. In these situations, physicians may diagnose a symptomatic individual through the use of a genetic test. That genetic information could then be used by insurance companies because it indicates an individual currently has a condition. However, if the patient’s children or other relatives of the patient elect to have presymptomatic genetic testing, the results of that test would be confidential because now there would be no diagnosis without clinical symptoms.

Expanded Newborn Screening

The 92nd legislative session enacted PA 92-0701 on July 19, 2002, which revises the Phenylketonuria Testing Act of 1965. It requires expanded newborn screening using tandem mass spectrometry. A separate effort in the House (HB 5871) to allow parents, guardians, and health professionals the ability to choose between the state and other private, accredited laboratories for the screening was not successful, and all newborn screens are still performed by the IDPH laboratory in Chicago.

Licensing of Genetic Counselors

The 93rd General Assembly enacted the Genetic Counselor Licensing Act as PA 93-1041. With the passage of this bill, Illinois became the third state in the nation to license genetic counselors. This legislation defines genetic counseling and requires anyone advertising his or her services as “genetic counseling” to be licensed. Licensing requires successful completion of the American Board of Genetic Counseling (ABGC) exam or the American Board of Medical Genetics (ABMG) exam. Temporary licenses are granted to recent genetic counseling academic program graduates who have not yet taken the exam, provided their work is overseen by a qualified supervisor. Furthermore, the legislation mandates that all information gained in genetic counseling sessions is considered privileged and confidential information and cannot be disclosed to outside sources.

Senate Bill 2012, submitted during the current 94th session, has passed both the House and the Senate and currently awaits action by the governor. This bill removes requirements in the Genetic Counselor Licensing Act that requires referrals for genetic counseling be written. If signed, this bill would allow physicians and other qualified health professionals to make oral referrals for genetic counseling. Further, genetic counseling would be permissible without any referral in some research settings.

Human Cloning Acts

Every legislative session, since at least the 90th General Assembly, has included some attempt to prohibit human cloning under the name “Human Cloning Act.” These bills all have in common the prohibition against cloning of the human ovum, zygote, embryo, or fetus as well as the purchasing and selling of the same for the purpose of cloning. The definition of “clone” is narrow enough so that only attempts to create human beings are included. The duplication of human DNA, tissues, organs, and cells would remain permissible. Several of these bills explicitly legalize therapeutic genetic cloning, provided no attempts are made at cloning an actual human being. Criminal and civil penalties would be established for violation.

The Senate passed these bills during the 91st and 92nd legislative sessions (SB 649 and SB 493, respectively), but no such bill has cleared the House. Beginning in the 93rd General Assembly, attempts were made to create a tandem Human Cloning and Adult Stem Cell Research Act (HB 6693 and SB 2934) that prohibits human cloning and simultaneously encourages research
using human adult stem cells. Criminal penalties for violations were lowered from Class 4 felonies to Class B misdemeanors; however, neither the Senate nor the House bill passed during the 93rd session. It has been resubmitted for the 94th session, with the criminal penalty for violation increased to a Class A misdemeanor. Additionally, language has been inserted into currently pending legislation (HB 3185 and SB 2100, see below) that would prohibit state funding of human cloning activities without broadly prohibiting those activities.

Stem Cell Research

During the spring session of the current, 94th General Assembly, several bills were proposed that would establish a mechanism of state funding for stem cell research (HB 3815, HB 2249, and SB 2100). The bills called for the establishment of an Illinois Regenerative Medicine Institute (IRMI) within the Department of Public Health that would oversee public support for research into adult, cord blood, and embryonic stem cells. Funding would come in part from a new 6% state tax on cosmetic surgery. Additionally, HB 3815 and SB 2100 propose that the 2006 general election ballot include a proposal a state issue of $1 billion in bonds to support stem cell research. However, none of these bills are expected to emerge from committee for voting during the 94th session and are therefore effectively defeated.

In mid July 2005, the Governor of Illinois, Rod Blagojevich, signed an executive order establishing the IRMI within the Department of Public Health. The order mandates $10 million in state funding be made available for scientific researchers investigating therapeutic uses of adult, cord blood, and embryonic stem cells. With the order, Illinois became the fourth state in the nation to provide funding for stem cell research.

SB 2002 was introduced by State Senator Bill Brady (R-44th District) and proposes that the IDPH develop a program for private storage of cord blood for individuals who cannot afford private blood bank storage. It would further require the IDPH to provide for the donation of stem cells at the time of an individual's death. This bill currently awaits action in committee.

Legislative Resolutions

Resolutions can be passed by either legislative house or by both houses in tandem. They are not signed by the governor and thus do not function as state law. However, they can provide insight into the lawmaker's strategies and plans for future legislation. The Illinois General Assembly has passed two such resolutions in the last five years.

The senate passed a resolution (SR 0292) in 2000 urging the University of Illinois to issue a report to the General Assembly on the medical, religious, biotechnological, genetics, legal, and bioethical consequences of human cloning. Although the resolution set a completion date of January 9, 2001 for the report, resolutions provide no funding mechanisms and the report was never completed.

The house adopted a resolution (HR 299) in late May that calls attention to Sickle Cell Disease and urges all public and private entities to inform the public of the effects of Sickle Cell on Illinois citizens. No specific programmatic activities were proposed.

Conclusion

The state General Assembly has targeted several major genetics issues over the past several years, and supporters have generally been successful in enacting legislation. This activity demonstrates the General Assembly’s willingness to consider important issues in the field.
Table 8.1 Review of Recent Major Medical Genetics Related Legislation in Illinois

<table>
<thead>
<tr>
<th>Session</th>
<th>Bill/Resolution</th>
<th>Status</th>
<th>Highlights</th>
</tr>
</thead>
</table>
| 90th General Assembly | HB 8 “Genetic Information Privacy Act” | Enacted as PA 90-0025 June 1997 | • Genetic testing and information derived from genetic testing is confidential and may be released only to the individual tested or his/her authorized representative.  
• Insurers cannot seek information derived from genetic testing for accident and health insurance. Individuals may submit genetic information to insurers, but it may only be used in policy decisions if the results are favorable to the individual.  
• No one may disclose or be forced to disclose the identity of any person upon whom a genetic test has been performed or the result of a genetic test that permits identification of an individual. |
| 91st General Assembly | SR 292                           | Adopted April 2000            | • Urges the University of Illinois to issue a report to the General Assembly on the medical, religious, biotechnological, genetic, legal, and bioethical consequences of human cloning.  
• Report will consider opinions of the general public.  
• Report will include a review and evaluation of current and past public policy and research related to human genetics  
• Target completion date of Jan 9, 2001; but study was never funded and not completed. |
| 91st General Assembly | SB 649 “Human Cloning Act”        | Passed Senate; died in House   | • Cloning defined as the creation or an attempt to create a human being.  
• Prohibits human cloning and the purchase or sale of an ovum, zygote, embryo, or fetus for the purpose of cloning a human being  
• Establishes that the penalty for violations is a Class 4 felony.  
• Explicitly refrains from restricting biomedical research from cloning human genes, cells, and tissues. |
<p>| 91st General Assembly | HB 631                           | Enacted as PA 91-0417          | • Provides that the Illinois Adoption Registry will serve as a mechanism for the voluntary exchange of genetic medical history between adopted children and their birth families. |
| 91st General Assembly | HB 1348                          | Enacted as PA 91-0549 August 1999 | • Updates state insurance code to reflect passage of PA 90-0025; limits insurers’ use of genetic information in the absence of a diagnosis related to that genetic information. |
| 92nd General Assembly | SB 493 / HB 3693 “Human Cloning Act” | Passed Senate; Died in House | • Substantively similar to SB 649 from 91st session; in addition to selling or purchasing human biological material for the purpose of cloning, adds to the list of restricted activities the transport or use of human biological material for the purpose of cloning. |
| 92nd General Assembly | HB 5870                          | Enacted as PA 92-0701 July     | • Amends Phenylketonuria Testing Act of 1965 to mandates expanded newborn screening for all infants through state NBS lab beginning July 1 2002. |
| 92nd General Assembly | HB 5871                          | Died in House                 | • Allows parents, guardians, and health care providers attending newborns to have choice of state or other accredited labs for tandem mass spectrometry newborn screens. All newborn screens currently conducted by state lab. |
| 92nd General Assembly | SB 42                            | Enacted as PA 92-0430 August 2001 | • Amends Genetic Information Privacy Act such that insurers that receive information derived from genetic testing may not use that information for a nontherapeutic purpose “regardless of the source of that information.” |</p>
<table>
<thead>
<tr>
<th>Sess-ion</th>
<th>Bill/Resolution</th>
<th>Status</th>
<th>Highlights</th>
</tr>
</thead>
</table>
| Illinois Genetic Services Needs Assessment | HB 4200 “Genetic Counselor Licensing Act” | Enacted as 93-1041 September 2004 | • Requires all health professionals who advertise themselves as genetic counselors to be licensed by the state.  
• Does not prevent others from engaging in this activity so long as they do not represent themselves as genetic counselors.  
• Regulates genetic counseling referral process.  
• Requires successful completion of the American Board of Medical Genetics or American Board of Genetic Counseling exam to be licensed.  
• Establishes temporary licenses for applicants who have successfully completed accredited genetic counseling training programs but have not completed the exam.  
• Temporary licensees may practice only under a qualified supervisor.  
• Genetic counseling information is designated as privileged and confidential. |
| 93rd General Assembly | HB 253 “Human Cloning Act” | Died in House                     | • Substantively equal to SB 649 from 91st session.                                                                                                                                                   |
| 93rd General Assembly | HB 3589 “Stem Cell Research Act”       | Passed House; Died in Senate      | • Expressly permits and encourages research involving the derivation and use of human embryonic stem cells, human embryonic germ cells, and human adult stem cells.  
• Requires that individuals receiving fertility treatment be permitted to choose between methods of disposing of embryos.  
• Individuals may not purchase or sell embryonic or cadaveric fetal tissue for research purposes.  
• Reimbursement for removal, processing, preservation, etc., is not considered purchase or sale. Likewise, embryonic or cadaveric fetal tissue may be donated. |
<p>| 94th General Assembly | HB 4201                                | Died in House                    | • Amends Genetic Information Privacy Act to require waivers of individuals' health history used by insurance companies to state that information derived from genetic testing is not included in the waiver. |
| 94th General Assembly | HB6693 / SB2934 “Human Cloning and Adult Stem Cell Research Act” | Both bills died in respective houses | • Combines substance of HB 253 and HB 3589 from 93rd session; lowers criminal penalty to Class B Misdemeanor |
| 94th General Assembly | SB 188 “Human Cloning and Adult Stem Cell Research Act” | Pending                          | • Resubmits substance of HB 6693 / SB 2934 from 93rd session; criminal penalty strengthened to Class A Misdemeanor |</p>
<table>
<thead>
<tr>
<th>Sess-ion</th>
<th>Bill/Resolution</th>
<th>Status</th>
<th>Highlights</th>
</tr>
</thead>
</table>
|          | HB 2249 / HB 3815 / SB 2100 “Illinois Regenerative Medicine Institute Act” | Pending* | • Requires that the 2006 general election ballot include the proposition that the state issue $1,000,000,000 in bonds to support stem cell research.  
• Research grants and loans will be overseen by the Illinois Regenerative Medicine Institute (IMRI).  
• Allows the Institute Oversight Committee and advisory committee to operate closed meetings.  
• Imposes tax of 6% on cosmetic surgery; 5% or $1,000,000 max of this tax goes into the IMRI Operations Fund.  
• Prohibits the state from funding research into human cloning. |
|          | SB 2002 | Pending | • Requires the IDPH to develop a program for providing private storage of cord blood for individuals who are unable to afford private cord blood banks.  
• Requires the IDPH to provide for the donation of stem cells at the time of an individual's death. |
|          | HR 299 | Adopted May 2005 | • Calls attention to Sickle Cell Disease and urges all public and private entities to inform the public of the effects of Sickle Cell on Illinois citizens. |
|          | SB 2012 | Passed both House and Senate May 2005; currently awaits governor’s action | • Amends Genetic Counselor Licensing Act to remove requirements that genetic counseling referrals be written.  
• Allows genetic counseling to be performed without referrals in some research settings. |
| Executive Orders | July 12, 2005 | n/a | • Establishes the Illinois Regenerative Medicine Institute within the Department of Public Health. Makes $10 million available for scientific research on therapeutic uses of stem cells. |

* Not expected to emerge from committee; effectively defeated
Chapter 9: Discussion and Next Steps

The purpose of this needs assessment is to assess the role and function of the state health department with regard to the current and future needs for genetic policy, service provision, workforce and community education, as well as research and surveillance in Illinois. This was accomplished through:

- An assessment of the current provision of genetic services in Illinois; and,
- An assessment of the essential elements of a state genetics program, currently and in the coming years, defined through the perspectives of key stakeholders in Illinois, and the visions and practices of key state and national public health agencies.

The field of genetics is experiencing rapid growth such that the context of this needs assessment was and is extremely dynamic. The needs, constituencies, available information and services are not only increasing exponentially but are doing so in a climate of uncertainty about how the field of genetics and health care in general will be affected. The goal is to assure that the expansion in genetics and associated shifts in medical care will ultimately benefit the health and well being of all Illinoisans as effectively as possible.

The survey and interview findings identified needs and barriers within public health and genetics, as well as perspectives about the role of the IDPH. The responses of genetic service providers result in a fairly strong consensus on the policy, education, and access issues that must be addressed while also expressing some uncertainty about how to resolve these issues. This reflects larger concerns about how genetics - as a field and as a constellation of clinical practices - should be defined, understood, and integrated into the larger health care system, and what an effective system of integrated genetic services will look like.

In this chapter we discuss key concerns and issues raised by participants within the categories of:

- Education, Information, and Engagement – for non-genetic health care providers, local health department staff, general public and patients, genetic service providers, state agency personnel, and advocacy workers;
- Genetic Service Provider Workforce – size and composition, distribution, role of genetic counselors, and future of the field;
- Reimbursement/Financial Coverage;
- Access Barriers;
- Ethical, Legal and Social Issues (ELSI);
- Programmatic and Data Issues; and
- Role of the IDPH Genetics and Newborn Screening Program.

This chapter ends with a schematic outlining several core issues and their associated influences and effects, which will be further developed during the 2006 planning process.

Education, Information, and Engagement

The need for additional education, information, and engagement was identified by and/or for all of the following stakeholders: primary care providers (pediatricians, family practitioners, internists), other non-genetic health care providers (specialist physicians, nurses, others), local health department staff, the general public and patients, advocacy workers, state agency personnel, and genetic service providers. The low interview response rate of the ‘other health care providers’
and the consumer/advocacy group representatives may itself indicate a need for education about genetics.

Educational needs identified by participants include general knowledge about genetics, identifying and referring patients for needed and appropriate services, and effective use of family history tools. These needs can be addressed, depending on the target audience, through conferences, lectures, continuing medical education, health profession school curricula, and school-based initiatives. Brochures, pamphlets, web sites, and resource directories are examples of identified informational needs.

Engagement involves stimulating interest in the topic of genetics and providing opportunities for dialogue." Engagement, or lack thereof, was not a dominant theme among survey respondents and interview participants; rather it was evident through the limited participation of some groups and the attitudes and input of others. Lack of engagement can be the result of the absence of knowledge, limited or no interest, lack of appreciation for the value of the issue, or competing demands. Evidence of each of these emerged from this needs assessment. Without engagement, it will be difficult for educational efforts to be effective.

The different types of needs suggest different educational strategies, which will be explored further in the development of the state public health genetic services plan.

Primary care and other non-genetic health care providers

The need to engage, inform, and educate primary care and other non-genetic health care providers (e.g., specialist physicians, nurses) was one of the most commonly identified needs and was mentioned by all types of interview and survey participants. The specific needs in this category included generating professional interest in genetics; addressing how to incorporate genetics into current practice; providing information about genetic referral resources; and basic and continuing education about genetics, identification of patients in need of a genetic services referral, use of family history tools, and ethical, legal, and social issues.

A local physician summarized several concerns as follows:

Some discoveries are so new we don’t know how to use them yet in our practice. You have a generation of physicians in practice who only learned simple genetics and haven’t been able to keep up with some of the more complex advancements. It also takes time to gather a complete family history of disease and know what to do with it. Genetic counselors are in short supply and we’re going to need more of them to help us understand how to interpret risk and what to do with it."

Some stakeholders felt that the issue of physician attitudes (e.g., about the importance and value of genetic services) could also be addressed through education. There was recognition that it is difficult and perhaps unrealistic to expect non-genetic health care providers to keep current with all new developments, as clinical genetic service providers acknowledged the difficulty doing so themselves. In addition, both genetic and non-genetic providers indicated that primary care providers have demands that compete with their ability to become engaged in genetics and that

---

www.cesagen.lancs.ac.uk/resources/docs/nowgenreview3.doc

time, knowledge and reimbursement constraints affect further integration of genetics into their practices.

Many genetic service providers expressed concerns about the competency of the larger health workforce and the adequacy of the genetics workforce in the state to meet current and future demands for genetic services. Our findings suggest that the knowledge needs of the health workforce can be addressed by:

- clarifying the professional qualifications necessary for the provision of specific services;
- determining how different medical fields and specialties can and will be organized to ensure an integrated and comprehensive system of care;
- identifying effective models of care;
- defining quality in the context of genetic service provision; and
- determining what a sufficient number of genetics professionals is and will be, in what contexts, and with respect to what models of care.

In considering the future of genetics, providers consistently emphasized the immense expansion in the clinical utility of genetics to a wide range of health concerns, including cancer and a number of common chronic diseases. They also noted that as genetics becomes an increasingly central aspect of all health care, the number and range of providers who will need to be able to effectively integrate genetics into clinical practice will increase. This was expected to be particularly true for primary care providers, who are already being called upon to order and interpret genetic tests, and some of whom are, according to genetic specialists, potentially overlooking important indicators of genetic risk that might alter the clinical course of treatment. Our findings suggest that the following questions guide the state’s response to these issues:

- What is important for providers in each profession and specialty to know and to be able to do?
- What is the most effective way to improve competencies within each field and overall? What role should the state play? What role should professional education institutions play? What role should professional associations play?
- By what standards of care should genetic services be provided in various settings in Illinois?
- What are the practical implications of recently enacted Illinois genetic counselors’ licensure requirements?

Genomics competencies for health care providers have been defined at the national level. For example, in 2001 (and updated in 2005) the National Coalition for Health Professional Education in Genetics (NCHPEG) defined “core competencies” in genetics for all health professionals. NCHPEG asserts that:

Each health-care professional should at a minimum be able to:

- appreciate limitations of his or her genetics expertise,
- understand the social and psychological implications of genetic services,
- know how and when to make a referral to a genetics professional.

225 Resource links include: American Academy of Family Practice (AAFP) Annual Clinical Focus, 2005-Genomics CME Video Series (www.aafp.org/x25023.xml); University of Kansas Medical Center Genetic Education Center’s Genetic and Rare Conditions web site (http://www.kumc.edu/gec/support/); National Human Genome Research Institute (NHGRI) operates the Genetic and Rare Diseases Information Center (GARD); http://www.genome.gov/10000409); March of Dimes Genetics & Your Practice site (http://www.marchofdimes.com/gyponline/index.bm); Resource links include: American Academy of Family Practice (AAFP) Annual Clinical Focus, 2005-Genomics CME Video Series (www.aafp.org/x25023.xml); University of Kansas Medical Center Genetic Education Center’s Genetic and Rare Conditions web site (http://www.kumc.edu/gec/support/); National Human Genome Research Institute (NHGRI) operates the Genetic and Rare Diseases Information Center (GARD); http://www.genome.gov/10000409); March of Dimes Genetics & Your Practice site (http://www.marchofdimes.com/gyponline/index.bm).
Implementing and refining these competencies for primary care will necessitate considering the heterogeneity within this field. This means recognizing and defining the different and relative roles, specific knowledge and skills base of pediatricians, family practice physicians, gynecologists, and other specialists within and across the health system. Addressing the informational and educational needs of the health workforce will necessitate clarifying the roles of key players including public health, medicine, and health professional education, and defining the tasks most effectively performed by each. Ensuring that the health workforce in Illinois is adequately prepared and capable of translating the scientific advances in medical genetics into actual benefits for patient health is best achieved in a collaborative effort of all stakeholders in the state.

Finally, the toughest challenge is not necessarily educating health care providers but engaging them in the issues and assisting them in applying their knowledge of genetics to clinical practice. Our findings suggest that effective collaborations would include health professional associations and educational institutions.

Local health department staff

“Genomics is helping us understand the role of genetic factors in leading causes of morbidity in the United States, information that public health professionals must be familiar with to improve health.”

Local health department staff expressed a need for more genetics education and training in such areas as identifying patients in need of a referral for genetic services and the most effective methods of referring patients to or delivering genetic services (including using the IDPH genetic screening tool and obtaining family/medical history). They also shared concerns about the adequacy of overall health department staffing in the context of integrating genetics into their programs. Health department staff have a variety of roles and responsibilities, especially those working in smaller, more rural health departments. The survey findings show that respondents have a low comfort level with providing a variety of services; this can serve as an indication of specific areas where training and education are needed. Also, while there were many compliments about the annual IDPH nursing genetics conference, there were also many requests for more frequent, shorter opportunities for training and continuing education at the local level. It is also important to generate interest in genetics from health department leadership and staff in all program areas; provide information about genetic referral resources and the effective use of genetic screening and family history tools; and provide education about genetics and related ethical, legal, and social issues.

Existing resources for training local health department staff include NCHPEG resources, the CDC Genomic Competencies for the Public Health Workforce (http://www.cdc.gov/genomics/training/competencies/comps.htm), and the Institute of Medicine’s publication: *Who Will Keep the Public Healthy? Educating Public Health Professionals for the 21st Century*.

General public and patients

The need to inform and educate the public was mentioned consistently throughout the interviews and surveys. With media coverage about genetic discoveries becoming more common, patients are asking their providers more questions about what they read and hear. Despite this trend, a lack of awareness and understanding of contemporary genetics persists among much of the

---

general public. This includes information about the value of genetic services, the interaction of genetics with lifestyle and behavior, how and where to obtain genetic services (including support services if an individual or a family member is diagnosed with a genetic condition), and the importance of knowing the family history. Misconceptions about genetics, and concerns about privacy and insurance and employment discrimination, will be essential components of any public awareness campaign, public service announcements, and/or school curricula.

There are existing resources for public education, including education in primary and secondary school settings. Some of the previously mentioned web sites have resources for public and patient education. The National Human Genome Research Institute (www.genome.gov) has educational resources for teachers as well as suggestions for scientists speaking to schoolchildren. The University of Kansas Medical Center Genetic Education Center (http://www.kumc.edu/geom) also has links to many resources for educators.

Deficits in public knowledge and awareness of genetics pose obvious concerns with respect to the public’s effective and appropriate utilization of genetic services. In the context of increased direct-to-consumer marketing and availability of genetic tests, deficits in public knowledge also pose concerns about the nature and meaning of informed consent and the potential for genetics-related harms. Ensuring an informed public will be an important aspect of consumer and public protection. Careful attention must be paid to defining the nature and scope of any public education effort. Our findings suggest that the following questions guide the state’s response to public education:

- What performance standards are needed for professional, institutional, commercial, consumer, public and other education about genetics?
- What are the most effective ways to reach and engage diverse communities to identify and understand their unique educational needs and address their potentially diverse concerns?
- What is the scope of concerns - insurance or employment discrimination, privacy, racism, eugenics, others?
- What are the best interventions to address public and professional misperceptions? Which beliefs are misperceptions, and which are deeper ethical and social concerns that need to be addressed by culturally competent systems of care and reforms in professional education and practice?
- What are the key implications for the public regarding ethical, social, and legal issues related to the expansion of genetic services?

Genetic service providers

Insight into the educational needs and interests of genetic service providers was obtained through both survey and interview responses. According to our survey findings, 47% of genetic counselors had been in their current work setting for five or fewer years and 61% of medical geneticists had been in their current work setting for 16 or more years. Different continuing education strategies will be required for those who are new to the field, have been in practice for a long time, or have specialized in a certain area. Findings from a national study of medical geneticists indicate that practicing geneticists “find it almost impossible to keep up with the knowledge explosion in their specific area … much [less] beyond their area(s) of interest.” In addition, there may be a need for genetic service providers to learn more about primary care and public health models of health care delivery. Another important role for genetic service providers

will be in facilitating the engagement of other health professionals and the public in the area of genetics.

Two advocacy participants spoke about the need for genetic service providers (or other diagnosticians) to be better informed about support organizations and to show more sensitivity in thinking about the experience of their patients. One of these shared considerable criticism of genetic and non-genetic health care providers, saying that they all need to show more sensitivity and support to families.

**State agency staff**

The reality of and potential for the role genetics can play in areas beyond newborn screening has been recognized by national professional associations and federal agencies, as well as locally by IDPH through the formation of the Genomics and Chronic Disease Prevention Team. This Team includes the leadership from IDPH’s genetics and chronic disease programs and was described in Chapter 3. Participation in this Team appears to be one important area in which IDPH staff obtained education and increased their awareness and understanding of public health genetics. However, application of genetics into other public health program areas is in its infancy. The CDC’s *Genomic Competencies for the Public Health Workforce* ([www.cdc.gov/genomics/training/competencies/comps.htm](http://www.cdc.gov/genomics/training/competencies/comps.htm)) includes guidance for public health professionals, including leaders and administrators, relevant to genetics/genomics.

State agency interviewees recognized the need to first educate themselves about public health and genetics before they can educate the public or agency leadership. The limited integration of genetics into other program areas also points out a need to strategize about how to engage public health staff and leaders. One discussion in the literature of genetics and public health noted, “public health professionals are essential to translating the capabilities of genetic tests into better population-wide health through their ability to promote research, treatment, and education about the interaction between environment, behavior, and genes.**”

There are also opportunities for staff from different program areas to benefit from knowledge about currently existing resources. For example, a suggestion arose to have genetic referral resources and other information available through regional Early Intervention (EI) offices to benefit those EI clients who have genetic conditions.

**Advocacy workers**

The advocacy interview respondents were a small and diverse group with different experiences, constituencies, and needs. This group had a low participation rate for interviews, with one non-participant indicating that “genetics is not even on our radar screen.” One of the advocacy group representatives suggested that it would be helpful for her and other advocacy workers to have a better understanding of what genetic services are and where they are available – a “one-stop” resource, perhaps via the Internet. She further suggested that each organization have an “expert” who knows how to help their constituents gain access genetic resources and who can answer general genetics-related questions.

**Genetic Service Provider Workforce**

There are a number of issues relevant to the genetic provider workforce that were identified as part of this needs assessment, including workforce size and composition; distribution; the role of genetic counselors; and the future of the field of medical genetics. Through our survey methodology, we identified 150 genetic service providers in Illinois. Out of these, 72 were genetic counselors, 42 MD or MD/PhD geneticists, 18 PhD (laboratory) geneticists, and 18 others, such as genetic nurses and MS/MD/PhDs not formally trained in genetics but by practice, position, or self-designation centrally engaged in clinical genetic service provision in Illinois.

Size and composition

Many participants in this needs assessment expressed the belief that there is an inadequate current and predicted future supply of geneticists and genetic counselors. There is currently one active residency program in the state for subspecialty training in medical genetics for physicians, sponsored jointly by the University of Chicago and Northwestern University. Northwestern University also has the only genetic counseling graduate program in Illinois, from which there were eight graduates in 2005, and a total of 35 graduates since 2001. Concerns about the number of clinical geneticists were voiced in relation to whether there will be enough to keep up with the increasing numbers of genetic tests available and advances in genetics knowledge, serve as a resource to primary care and other health care providers, and work with genetic counselors, who currently cannot bill on their own for services.

Recent publications echo these supply concerns at the national level. Nationally, the number of individuals who achieve board certification in medical genetics each year has declined markedly since 1982. Authors of a national study on the geneticist workforce concluded, “There is a serious mismatch between the expansion of knowledge and clinical applications in the field of medical genetics and the size of the medical genetics workforce…. The obvious question is why has the profession not grown and attracted new entrants?” For further discussion of this issue, see below, Future of the Field.

Our survey results, also supported by national data, indicate a lack of racial and ethnic diversity among genetic service providers. Ninety-three percent of genetic service provider respondents were white, non-Hispanic. The median age of medical geneticists in Illinois is 53 years, with 34% of respondents aged 55 years or older (approaching retirement age). Genetic counselors in Illinois, almost all women, are much younger than the geneticists, with a median age of 32 years. We know of no data on the workforce participation of genetic counselors during child-

---

230 RNs who practice in genetics and self-identify as genetics nurses via membership in ISONG, GTFI, or through listing on NSGC’s Find a Counselor Search.
231 Self-designated practitioners represent those that have included in their AMA Doctor Finder profiles ‘medical genetics’ as a self-designated specialty.
232 K. Ormond, personal communication.
rearing years, but other professions that are dominated by women, such as nursing, show a decline in workforce participation for married women with children.\textsuperscript{237}

**Distribution**

None of the genetic service providers identified through our survey methodology were located in non-metropolitan areas. Many survey and interview participants noted a lack of genetic service providers in their local areas, particularly in rural areas. Currently, IDPH attempts to address geographic distribution problems by having geneticists travel to other areas of the state to hold regularly scheduled (e.g., monthly, quarterly) outreach clinics. As one potential solution to the problem of maldistribution, we asked genetic service providers about telemedicine; 43\% of respondents reported that while they had not used telemedicine, they thought it might be an effective way to deliver services and another 10\% said they had used it and thought it was somewhat effective.

**Role of genetic counselors**

In 2004, Illinois passed the “Genetic Counselor Licensing Act,” which was strongly supported by Illinois genetic service providers through the Genetic Task Force of Illinois (GTFI). However, there is currently no mechanism for genetic counselors to bill or be reimbursed for their services. Since there are not a large number of genetic counselors, especially outside of large cities, there also is a concern about how the licensure requirement will impact the ability of non-licensed health care professionals, especially nurses, to provide genetic counseling services to rural patients. Another issue raised was the dependence of many genetic counselors on the IDPH grants for their salary support.

**Future of the field**

There is currently national debate and discussion about where the future of medical genetics lies, as articulated by the president of the American College of Medical Genetics\textsuperscript{238} and other leaders in the field.\textsuperscript{239} Likewise, Illinois genetic service providers had both strong opinions and questions about the future of genetics as a specialty. There was agreement that genetics-related knowledge is increasing exponentially, as is the availability and complexity of genetic tests. National data referred to above also show that the number of new entrants to the geneticist workforce is shrinking. This paradox leads to a number of questions about how genetic knowledge and services will be provided to an ever-growing number of people by fewer providers, including: whether primary care providers will bridge this gap by offering more genetic services; how they, or other, non-genetics providers will “get up to speed;” what primary care providers should do or be trained to do, what they have time for and can be reimbursed for; how practice delineation or specialization among geneticists will affect access to care; whether geneticists will become (as one interviewee suggested) more like gatekeepers to testing services and educators of other health care providers; and, with decreasing entrants, whether genetics can and will remain a medical specialty.


The attractiveness of the profession affects recruitment of new geneticists and the future of the field. The median net professional income of MD geneticists was $135,000 in 2002.\textsuperscript{240} In comparison, the mean physician income in 2000 was $205,700.\textsuperscript{241} At a genetics professional summit, 40\% of participants cited “low compensation for medical geneticists” as a reason for the declining number of physicians seeking medical genetics training.\textsuperscript{242} Most genetics departments lose money, since reimbursement is limited or nonexistent for many services. Reimbursement for services provided is considered a critical issue for the future of the profession.

**Reimbursement/Financial Coverage**

Genetic service providers reported that genetics programs/departments often do not cover their costs and are therefore supported by the hospital or academic medical center, as much of what genetic service providers do is not billable or reimbursable, such as researching genetic conditions to reach a diagnosis, securing pre-approval, and obtaining informed consent. Also, as noted, genetic counselors currently cannot bill for their services.

A majority of genetic service providers identified insufficient and uneven reimbursement policies as a key constraint on effective and equitable service provision. The coverage policies under Medicaid were considered especially problematic given the disparities they create in access for a disadvantaged population. Related to this issue is the concern expressed by many providers about the lack of mechanisms (e.g., CPT procedure codes or ICD-10 diagnostic or procedure codes) for reimbursement for clinical services that are considered to be essential components of quality genetics care. While genetic counseling was most frequently identified in this regard, intellectual time, or time spent researching to determine diagnoses or appropriate care interventions, was also noted.

Aside from the strain on providers, and the systemic instability of care in the context of uncertain reimbursement, these issues were noted to have implications for access to and quality of care. Both genetic and non-genetic health care providers expressed concerns about cuts in state funding for genetic services. Genetic service providers reported that a patient’s insurance coverage – or willingness to pay out-of-pocket – affects his/her decisions about genetic testing and other services. One geneticist stated: “I fear ‘genetics for the wealthy’.”

A key issue is assuring that patients are adequately informed of the potential social, legal, ethical, and personal consequences of decisions regarding genetic screening. However, if counseling is not a service that can be reimbursed, then this important and time consuming component of care will most likely not occur outside of the specialized settings in which genetic counselors currently work. This has important implications for considering both the quality and ethics of care within the genetic services system and will require defining the following more clearly:

- the essential elements or necessary components of quality genetic services;
- standards of care with respect to genetic service provision across different specialties, subspecialties and contexts of care (i.e., prenatal, pediatric, adult);
- the meaning of a billable unit of time;
- the mechanisms necessary to ensure that all essential services can be reimbursed;


• the minimum set of genetics related services that all insurers should include in standard coverage; and,
• how equitable standards of coverage will be ensured across different insurers.

Interviews with non-genetic health care providers revealed financial and reimbursement issues were an issue for them as well. One provider noted that he could not afford the time and hassle required to write letters of medical necessity to insurance companies to assist patients in securing coverage. Participants also reported limitations on reimbursement from insurance companies that restrict the ability of primary care providers to become involved in genetics.

Access Barriers

Participants identified three main disparities which affect access to genetics care: geographical disparities which create barriers for those residents living outside the Chicago or other metropolitan areas; disparities in service utilization determined by insurance status and coverage; and disparities due to the absence of linguistic and cultural diversity/competency among genetic service providers. Another issue, raised mainly by local health department staff, was patient non-compliance and limited education, as well as the problem of missed appointments. These barriers are similar to those raised for other specialty areas of medicine.

Assuring access was identified by a number of genetic service providers as a key role for the state, as was direct service provision, to guarantee access to services that would not otherwise be provided or received. A number of questions remain about what this means and how best to accomplish a more equitable system of service provision. These questions include:

• What is the most effective way to provide access to services in areas of the state where the level of demand may not support locating genetic specialists? How will this change as primary care providers become increasingly important participants in genetic services?
• Should access to support services also be considered? What defines the scope of what services are needed and not available in non-metropolitan counties?
• How can the state ensure that well documented disparities in health by race/ethnicity and socioeconomic status are not exacerbated by the expansion of genetics? What might the different concerns of different communities be? Are all communities facing the same barriers in terms of access?

Ethical, Legal and Social Issues (ELSI)

Survey respondents and interview participants raised ethical, legal, and social issues that suggest a need for clear policy guidance concerning how, in the context of expanded testing options, especially pre-symptomatic testing, the public will be protected from such harms as genetic discrimination, the inappropriate use of personal genetic information, violations of privacy and breaches of confidentiality, and inappropriate testing with or without informed consent. While one aspect of such protection may occur through population-level education, there is a clear need for a comprehensive policy response to these issues as well. In considering this from a policy perspective, it will be important to identify whether and to what extent patients are currently experiencing genetic discrimination and/or other harms, to assess how well current regulations are protecting patients from such harms, and to determine what laws and regulations will need to be in place to ensure and assure that research and service expansion in genomics will do more good than harm in Illinois.
Programmatic and Data Issues

From a review of secondary data and conversations with key stakeholders, there appears to be a need for improvement in the completeness, usefulness, quality, and accessibility of state datasets, as well as in the ability to link them. There is currently recognition on the part of IDPH of the need to upgrade the data systems for greater integration and linkage capabilities. Other, specific suggestions from participants included the need to collect data about the cost-effectiveness of genetic services, evaluate the private and public caseloads of funded regional genetics centers, develop a resource database, gather data for gene-environment interactions, and provide data on the incidence of specific genetic conditions.

Role of IDPH Genetics and Newborn Screening Program

This section describes participants’ perspectives on the role of IDPH’s Genetics and Newborn Screening Program in the Illinois genetic service system. Some questions to consider when examining the list below and thinking about the needs and issues identified above are:

- What is the most appropriate way to orient the program within a context of finite, limited resources?
- What are the strengths of the current program? What are the weaknesses?
- Should the program’s focus be expanded? If so, to what areas and how?
- Which identified needs can be influenced at the state-level, and by a state agency, and which are national, systemic issues?
- What is feasible and appropriate for different stakeholders, including the IDPH Genetics and Newborn Screening program, to engage in or take leadership of? And, how can these different stakeholders be brought into the process?

These questions will be addressed in the next phase of the project, which is the development of a state public health genetic services plan.

Participants’ perspectives on the role of IDPH’s Genetics and Newborn Screening Program included:

- **Funding** – many participants felt that IDPH should provide funding for:
  - expanded genetic services and testing
  - grants for genetic counselor salaries and to local health departments (maintain and increase current grants)
  - local genetics resources and services
  - training and continuing education of genetic professionals
  - outreach and education

- **Education**:
  - public
  - local health department staff
  - non-genetics health care providers
  - genetics providers
  - an IDPH team to coordinate all of the above programs
• Leadership role on genetics-related coverage and reimbursement issues – educational/lobbying efforts to address the following needs:
  o improved Medicaid reimbursement rates
  o expanded coverage of genetic tests and services by Medicaid
  o increased coverage of genetic tests and services by private insurance companies
  o ability of genetic counselors to bill
  o service coverage for indigent populations

• Coordinate and integrate genetics services into general health care
  o use authority as state health agency to assure population access to genetic services
  o build stronger networks with national organizations
  o provide informational resources to genetic disease support organizations
  o streamline genetic referral processes

• Data and surveillance.
  o coordinate state databases
  o improve use and completion of current data

• Ethical, legal, and social issues (ELSI) – there is a need for policy guidance concerning
  o genetic discrimination
  o inappropriate use of personal genetic information
  o violations of privacy and breaches of confidentiality
  o inappropriate testing and testing without informed consent

Conclusions and Next Steps

The IDPH Genetics and Newborn Screening Program has had many accomplishments and its activities and staff received many compliments throughout this project. The program has an impressive infrastructure in place. Program administrators have taken a proactive approach in conducting this needs assessment to learn what key stakeholders in the state perceive as unmet needs, barriers to access, the future of genetic services delivery, and the role of IDPH. As genetics evolves, there are many opportunities for the state public health program to evolve, including, for example, broader population screening, health promotion and disease prevention activities, expanded access to and provision of services, workforce training, improved data infrastructure, public education, and research. While the opportunities are endless, the resources, of course, are not.

Figure 9.1 shows four “core issues” identified by needs assessment participants and in the national literature. They are particularly relevant in the context of rapid expansions in genetic knowledge, services, testing, and technology. The core issues are: limited genetic provider supply; low public genetic literacy; inadequate third-party reimbursement; and lack of integration between genetics and the overall health care system. This diagram is not an exhaustive list of core issues, or influences and effects, rather it illustrates several major themes that were raised throughout this process.

This needs assessment, by definition, identifies a range of interrelated problems, whose solutions will require various approaches. For instance, the supply shortage of genetic service professionals will impact access to and provision of genetic services in the state. Yet it is not the sole province, or even within the primary mission, of the Illinois Department of Public Health to increase the number of new geneticists. Instead, within the public health functions of assurance and
policy development, an appropriate role for the department might be to offer leadership for a collaborative response.

The overall challenge for IDPH with respect to the comprehensive field of genomics is to define the system in which the spectrum of issues, trends, and actors are and will be functioning, and then to set performance standards for this system. This approach to planning identifies leadership within the state, makes known the scope of activity the department can and should address, outlines what should be undertaken in partnership with other organizations or agencies, and sets accountability standards for these and other players within a 'genomics-competent' state.

The findings discussed in this report will serve as a basis for the next phase, the development of a public health genetic services plan for Illinois. That phase will involve stakeholders in Illinois in a systematic process of engagement, education and policy recommendations, as persons throughout the state develop a consensus about what needs and opportunities should be pursued, by IDPH and perhaps by other organizations, and how these should be prioritized.
Figure 9.1 Overview of Core Issues Related to the Provision of Genetic Services

**Influences**
- Attractiveness of field
- Limited pipeline programs
- Need for genetics education for non-genetic health care providers
- Need for education for genetics providers about primary care models
- Inadequate reimbursement and Insurance
- Lack of appreciation of and education about genetics by:  
  - public
  - payers
  - policy makers
- Complexity of genetics field and terminology

**Core Issues**
- Limited genetic provider supply
- Low public genetic literacy
- Inadequate 3rd party reimbursement
- Lack of integration between genetics and overall health care system

**Potential & Actual Effects**
- Inequities in access
- Inequities in quality
- Limited supply of genetics providers
- Inadequate pipeline
- Increasing provision of genetic services by non-genetic health care providers
- Need for consumer protections
- Need for information and education for non-genetics health care providers, public, policy makers, payers

**Additional Effects /Needs**
- Lower health outcomes
- Lack of ethnic diversity
- Lack of geographic diversity
- Education for non-genetic health care providers
- Delineation of roles for genetic/non-genetic providers
- Develop standards of care
Appendices
Appendix 1: Methods

The needs assessment process involved the review of key documents and program literature, as well as surveys and informant interviews with key stakeholders throughout the state. These stakeholders included policy makers, genetics and other health care providers, consumer and health advocacy groups, and personnel from state agencies and local health departments.

The Institutional Review Board (IRB) at the University of Illinois at Chicago approved this study, including the survey questionnaires and the interview guides.

Surveys

Questionnaire Development.

Questionnaires were developed for two different groups: (1) genetic service providers, defined as certified MD and PhD geneticists, clinical providers (MD, PhD, MS) with non-genetic certifications but practices that directly involve genetic service provision, genetic counselors, and genetic nurses; and (2) local health department personnel, representing genetic coordinators and IDPH newborn screening contacts. The questionnaires were developed after review of other available surveys. Both questionnaires were reviewed by IDPH personnel and selected genetics providers for appropriateness and feasibility.

The Survey for Clinical Genetics Service Providers (Appendix 3) consisted of 45 questions (12 pages) organized into the following sections: Provider Practice; Genetics Patient Care/Clinical Practice (only respondents who indicated that they provide “face-to-face” patient care); Clinical Genetics Laboratory Services (only respondents who indicated that they work in a clinical genetics laboratory setting or provide clinical genetic laboratory services); Perspective; and Provider Background.

The Local Health Department Survey (Appendix 2) consisted of 21 questions (5 pages) organized into the following sections: Background; Genetic Services (only respondents who indicated that their health department currently provides genetic services); and Genetics and Public Health.

Data Analysis

Data were entered into separate Excel spreadsheets for each survey. The Excel data were imported into SAS for statistical analysis. Most data are descriptive; variable definitions are, when necessary, provided in the survey findings sections. Many questions on the Survey for Clinical Genetics Service Providers asked respondents to select categorical responses that contained numeric values (e.g., from a range of percent values, such as 1-10%). For the categorical responses with numeric values, we present grouped mean values (unless indicated) rounded to the nearest whole number. The grouped mean values were calculated by multiplying the midpoint value of each category by the frequency of responses in each category, summing over all of the categories, then dividing by the total frequency.

243 Individuals with certification through the American Board of Medical Genetics in: Clinical Biochemical Genetics, Clinical Biochemical Molecular Genetics, Clinical Cytogenetics, Clinical Genetics (MD), Clinical Molecular Genetics, and/or PhD Medical Genetics.

244 Surveys from other states, including Michigan, Oregon, and Washington, as well as the 2003 Survey of ABMG Certified Geneticists, developed by JA Cooksey, et al. at the University of Maryland-Baltimore.
Sampling Frame and Response Rate: Survey for Clinical Genetics Service Providers

The sample of genetic service providers was developed based on website review, national and local organization directories, and consultation with IDPH personnel and key genetic providers in the state. The following web-based resources were reviewed in developing the sample: American Society of Human Genetics (ASHG) Genetics Societies Membership Directory; American Board of Medical Specialties (ABMS) Specialist Search; National Society of Genetic Counselors (NSGC) Find a Counselor Search; and the American Medical Association Doctor Finder. In addition, websites for genetic centers and departments at or affiliated with Evanston Northwestern Healthcare, Children’s Memorial Hospital, Rockford Health System, Northwestern University, University of Illinois (Chicago, Peoria, Urbana), Southern Illinois University, Rush University, University of Chicago, and Loyola University, were reviewed, along with websites from state and local genetic advocacy organizations. Membership directories from the International Society of Nurses in Genetics (ISONG) and the Genetic Task Force of Illinois (GTFI) were also utilized. To ensure completeness, selected genetics providers reviewed the final sample.

The original sample consisted of 155 providers, representing: genetic counselors (n=73), MD/PhDs certified in genetics245 (n=64), and others (n=18), such as genetic nurses246 and MS/MD/PhDs not certified in genetics but by practice, position, or self designation247 centrally engaged in clinical genetic service provision in Illinois. Surveys were mailed to all 155 providers on October 31, 2004. An e-mail was sent about the same time to all GTFI members letting them know they would receive the survey in the next few days and encouraging them to respond. Three surveys proved undeliverable in this initial mailing, resulting in a final sample of 152. Sixty-one complete surveys were received, for a response rate of 40%.248 Reminder postcards were mailed on November 30, 2004 to all individuals on the initial list excluding four individuals whose response status was known.

On February 4, 2005, the survey was resent to 148 individuals from the original sample, representing the original sample of 155 less seven individuals whose response status was known (three undeliverable, two declining participation, and two known to have completed the survey following the initial mailing). Twenty-four surveys were returned complete, of which seven were determined to be duplicates249 Two surveys were returned undeliverable.

After adjusting the sample size to 150 to account for the 5 undeliverable surveys, the overall response rate was 52% (78/150).

Of the revised sample of 150 genetic service providers, 72 (48%) were genetic counselors, 42 (28%) were MD or MD/PhD geneticists, 18 (12%) were PhD (laboratory) geneticists, and the remaining 18 (12%) were part of the previously described other category. All providers were located in metropolitan areas, with 81% located in Cook County. Seventy-seven percent of the providers were women; almost all (94%) genetic counselors, 41% of MD or MD/PhD geneticists, and three-fourths of PhD geneticists were women.

245 See footnote 243.
246 RNs who practice in genetics and self-identify as genetics nurses via membership in ISONG, GTFI, or through listing on NSGC’s Find a Counselor Search.
247 Self designated practitioners represent those that have included in their AMA Doctor Finder profiles ‘medical genetics’ as a self designated specialty.
248 Response rates were calculated in accordance with AAPOR Standard Definitions, on-line edition 3.1, Feb. 2005, utilizing formula RR1 for Minimum Response Rate. The AAPOR defines response rate as the number of completed surveys divided by the number of eligible units in a sample. The response rates calculated herein exclude from the calculation of eligible sample (a) undeliverable surveys (i.e., surveys returned unopened by the postal service) and (b) duplicates (this category is relevant only to the genetic provider survey).
249 All surveys from the second mailing were assessed for potential duplication. Duplicates were determined by matching demographic and professional information, responses, and handwriting. Duplicates were removed from the analysis.
Sampling Frame and Response Rate: Local Health Department Survey

We received a list of local health department contacts from IDPH. There are 94 local health departments in Illinois, 39 of which receive funding from IDPH for the provision of genetic follow-up and education services. Surveys were mailed to all 94 health departments, addressed to the Genetics Coordinator at the 39 funded health departments, and to IDPH’s contact for newborn screening follow up (most frequently representing the positions of Maternal Child Health Coordinator and/or Nursing Supervisor) at the unfunded health departments.

The initial mailing to all 94 health departments occurred on October 26, 2004. Reminder postcards were sent November 30, 2004. Reminder emails with attached surveys were sent to non-responding Genetic Coordinators on December 28, 2004 (n=16; survey response was determined by zip code provided in the completed surveys). A second mailing of the survey to all non-respondents (n=40) was sent February 4, 2005. Prior to the second mailing, 54 complete surveys were received for an initial response rate of 57%. Following the second mailing, the response increased to 75.5% (71/94).

Key Informant Interviews

Sample Selection

In depth interviews were planned with representatives of major stakeholder groups in the Illinois genetic service delivery system (see Figure 1). The target sample included genetic service provider representatives from all 16 state funded genetic centers, additional medical and lab geneticists and genetic counselors, representatives from state agencies, genetic or maternal and child health coordinators at local health departments, and other health care providers (medical specialists, primary care physicians, and nurses). For all relevant groups, care was taken to ensure adequate representation throughout Illinois.

Focus groups were originally planned with representatives from advocacy organizations. However, because of a low response to the focus group invitations, we instead invited these organizations to participate in-depth interviews. All organizations were invited to participate in interviews, even if they had declined to participate in a focus group.

The genetic services provider sample was first based on ensuring representation from each of the 16 state-funded regional genetic centers. The official contact representative at each center was asked to participate in an in-depth interview. Replacements were made for representatives who declined an interview or were unable to schedule a time. Other geneticists and genetic counselors were selected according to criteria that included geography and area of specialization.

The list for the other health care provider sample was developed through consultation with genetic providers. The types of medical specialists included were determined by knowledge of where genetics currently plays a role in diagnosis and treatment of patients, or where advances in genetics are expected to play a role within the next several years.

We chose to interview staff at 14 health departments, half with IDPH genetics grant funding and half without. In addition, we added geographic area to our criteria, based on the seven Illinois regions used by IDPH. Therefore, we selected two health departments from each of the seven IDPH regions, one “funded” and one “unfunded.” Selection was randomized to the greatest extent possible; randomization was constrained to exclude contiguous localities. Contact names for funded

---

250 This is a position specifically created and maintained by IDPH’s funding program; thus, only funded LHDs have ‘Genetic Coordinators.’
health departments were obtained from IDPH. For unfunded health departments, the invitation was sent to either the genetics coordinator, if applicable, or the MCH coordinator.

State agency representatives were targeted at the Illinois Departments of Public Health (IDPH), Human Services (IDHS), and Public Aid (IDPA), and the Department of Specialized Care for Children (DSCC). As with the other health care provider sample, individual state agencies were selected whose constituency currently consists of clients with genetic concerns or who are predicted to use genetics to a much greater extent over the next several years. Invitations were sent to the director or chief of the relevant program.

Finally, the advocacy groups were categorized three ways: (1) genetic disease/condition advocacy; (2) chronic disease advocacy; and (3) other health or ethnic advocacy. If the group was a national organization, then the state and local chapters were selected. Invitations were usually sent to the president or executive director of the organization, who could elect to delegate the interview or do it himself.

We added two practitioners of medical law and ethics to the target sample during the invitation process.

**Figure 1 The Target Sample**

**Invitation Methodology**

Invitation letters and an IRB stamp-approved consent form were usually both e-mailed and sent via regular mail to invitees. Both the e-mail and hard copy invitations included information for scheduling interviews by phone, email, or fax. Follow up phone calls were made to invitees to check on the status of the invitation at approximately one week intervals thereafter; messages were left on voice mails or with assistants if the invitee was not available at the time of the call. A maximum of three phone calls were made before the individual was considered to have passively declined the invitation. The date of invitation, follow up phone calls, and responses were all progressively logged throughout the duration of the invitation process.
Responses were categorized as accept, decline, passive, or ineligible. Passive refers to individuals that did not respond to the invitation or follow up phone calls. Ineligible refers to individuals who were deemed inappropriate for this study; for health care professionals, this was usually either because the invitee had relocated out of state or was otherwise no longer practicing the targeted discipline. For public health representatives, this was normally because the individual had been reassigned to a different agency and/or organization.

Once an invitation was accepted, an interview time convenient to the respondent was arranged; one of the members of the interview team was then assigned to conduct the interview. Most respondents were sent a confirmation of the interview time several days in advance; this confirmation included a copy of the interview tool.

**Response Rate**

Table 1 below summarizes the invitation process and response rate. Raw numbers are given for the size of the target sample, total invitations sent, and actual sample size. Actual sample size refers to the final sample; that is, the number of completed interviews used for analysis. The target rate is the ratio of the actual sample size to the target sample size. The response rate is the ratio of the sum of responses categorized as “accept” or “decline” to the difference of total invitees and responses categorized as “ineligible.”

**Table 1 Summary of Target and Actual Sample Responses**

<table>
<thead>
<tr>
<th>Group</th>
<th>Target Sample Size</th>
<th>Total Invitations Sent</th>
<th>Actual Sample Size</th>
<th>Target Rate (%)</th>
<th>Response Rate (%)</th>
</tr>
</thead>
<tbody>
<tr>
<td>16 Regional Genetic Centers</td>
<td>16</td>
<td>20</td>
<td>16</td>
<td>100</td>
<td>100</td>
</tr>
<tr>
<td>Other Genetic Providers</td>
<td>20</td>
<td>29</td>
<td>18</td>
<td>90</td>
<td>88</td>
</tr>
<tr>
<td>Other Health Care Providers</td>
<td>23</td>
<td>42</td>
<td>10</td>
<td>44</td>
<td>55</td>
</tr>
<tr>
<td>Local Health Departments</td>
<td>14</td>
<td>18</td>
<td>14</td>
<td>100</td>
<td>100</td>
</tr>
<tr>
<td>State Agencies</td>
<td>35</td>
<td>41</td>
<td>21</td>
<td>60</td>
<td>83</td>
</tr>
<tr>
<td>Advocacy</td>
<td>24</td>
<td>29</td>
<td>8</td>
<td>33</td>
<td>34</td>
</tr>
<tr>
<td>Total</td>
<td>132</td>
<td>179</td>
<td>86</td>
<td>65</td>
<td>72</td>
</tr>
</tbody>
</table>

Genetic practitioners were overall very receptive to interview invitations; the combined response rate of all genetic providers was 93%. Likewise, all representatives from local health departments responded to the invitation, with only a small number declining. The 100% target rate for local health department includes the sample goal of one funded and one unfunded department from each of the seven IDPH regions.
The lowest response and target rates come from the Advocacy and Other Health Care Provider samples. Only about one-third of the interviews targeted for these samples were completed. Four physicians who consented to interviews were nonetheless unable to schedule a convenient time. Primary care providers were especially difficult to recruit for this study. A total of 19 family practice physicians, internists, obstetricians, or primary care pediatricians were invited to participate and only two consented to interviews.

The Interview Process

Each separate sample had its own interview tool, which covered similar content areas although questions were customized to the particular targeted sample. Each guide consisted of approximately 10-15 questions; guides are included in Appendix 4. Interviews generally took 20-30 minutes. Most interviews were tape recorded per the interviewee’s consent; interview summaries were conducted using the taped conversation as a reference. Most interviews were conducted with just a single interviewer; at the start of the interviewing process, however, members of the interviewing team did interviews in pairs. Each interviewer wrote up the interviews for which they were responsible. For ease of analysis, summaries were quasi-transcriptional and captured the interviewee’s exact phrasing while rendering the speech in a prose-based style. Normal conversational feedback was not captured. Those interviews that were not tape-recorded were written up based on the interviewer’s notes.
Appendix 2: Local Health Department Survey

ILLINOIS STATEWIDE GENETICS NEEDS ASSESSMENT
Local Health Department Survey

Please fill in the blank or circle where appropriate.
THANK YOU for your participation!

I. Background

1. What is your current position? ______________________________________

2. What is your professional degree(s)? Select all that apply.
   O BA
   O BBA
   O BS
   O DO
   O Dr.P.H.
   O RN or BSN
   O MBA
   O MD
   O MPA
   O MPH
   O MS
   O MSW
   O PA
   O PD
   O MSN or Clinical Nurse Specialist
   O MSW
   O PA
   O RD

3. What year did you receive your most recent professional degree? _______

4. What is the zip code at your local health department? _________________

5. Does your health department currently provide genetic services?
   O Yes, CONTINUE with next question
   O No, SKIP to question 15

II. Genetic Services

6. Does your health department receive a grant from IDPH to provide genetic services? O Yes O No

7. Which of the following genetic services does your health department provide? Select all that apply:
   O Newborn screening follow up
   O Genetic screening, please indicate which screening tool you use:
   ____ IDPH Genetic Screening Tool
   ____ Cornerstone
   ____ Other
   O Onsite genetic clinic
   O Referral to genetic provider (MD/DO Geneticist, Certified Genetic Counselor) for genetic evaluation/screening.
   O Referral to physician (Primary Care, OBGYN, GP) for genetic evaluation/screening.
   O Public education on genetics
   O Professional education on genetics
   O Sample collection for genetic laboratory testing
   O Other, specify: ____________________________________________

Examples of genetic services include:
- Screening of children or adults for genetic disorders/conditions
- Follow up for positive newborn screens
- Client referral to genetic centers
- Educational presentations on genetics and health
8. Does your health department advertise these services?

O Yes           O No

8.1. If yes, to whom does your department advertise?
Select all that apply.

O MD/DOs       O Other Health Departments
O Nurse Practitioners O Community Health Centers
O Hospitals     O Genetic Centers
O Birthing Centers O Other, specify: _________
O Consumer Groups O General Public
O General Public

8.2. Briefly indicate how your department advertises (e.g., announcements mailed, adverts in local papers, information on website etc.):


9. What programs in your health department are currently involved in the provision of genetic services (e.g., NBS follow up, genetic screening, etc.)? Select all that apply.

O Genetics       O Doula       O Cardiovascular
O APORS        O Perinatal Care   O Cancer
O WIC            O AOK, 0-3      O Other, specify_________
O Family Case Management O Hearing/Vision Screening
O Family Planning O Diabetes        O Osteoporosis
O Teen Parent Services

10. Please indicate the number of full-time equivalent staff at your health department currently involved in the provision of genetic services.

# FTE

_________ Genetic Coordinator, please indicate degree
_________ Nurse (RN or higher)
_________ Case Manager
11. At your health department, what is the position and degree of the staff member(s) responsible for making referrals to genetic centers?

Position: ____________________________
Degree: ______________________________

O Don’t know
O Not Applicable

12. Do you feel that your department’s current provision of genetic services adequately meets your clients’ needs?

O Yes     O No       O Don’t Know

13. If you answered NO to #12 please indicate what needs you feel are currently NOT being met?

14. Please name the genetic center/clinic/practice your health department refers clients to most often: ____________________________________________

III. Genetics and Public Health

15. How much of an impact do you think recent advances in human genetics (e.g., the completion of the human genome project) are having and will have on your health department’s programs?

<table>
<thead>
<tr>
<th></th>
<th>None</th>
<th>Very Little</th>
<th>Some</th>
<th>A lot</th>
<th>Don’t Know</th>
</tr>
</thead>
<tbody>
<tr>
<td>Currently</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>In the next 5-10 years</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>In the next 10-15 years</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
</tbody>
</table>
16. In your opinion, what chronic disease programs will need to incorporate genetic information over the next 3-5 years? Select the top 3 priorities.

- O Asthma
- O Cancer
- O Cardiovascular Disease
- O Diabetes Mellitus
- O Alzheimer’s Disease
- O Obesity
- O Osteoporosis
- O Arthritis
- O Other ______________________

17. Are there any other public health program areas (aside from the chronic disease programs listed above) that you think will need to incorporate genetic information over the next 3-5 years?

- O Yes, please specify: ____________________________________
- O No
- O Don’t Know

18. How important do you think the following services are to your health department’s role and what is your level of comfort with the ability of your health department to provide each service?

<table>
<thead>
<tr>
<th>Role</th>
<th>Comfort</th>
</tr>
</thead>
<tbody>
<tr>
<td>a. Public/Patient Education About:</td>
<td></td>
</tr>
<tr>
<td>Inheritance patterns and principles</td>
<td>Very Important</td>
</tr>
<tr>
<td>Birth defect prevention strategies</td>
<td></td>
</tr>
<tr>
<td>Possible genetic risks to offspring.</td>
<td></td>
</tr>
<tr>
<td>Genetic risk in common chronic diseases.</td>
<td></td>
</tr>
<tr>
<td>Genetic risk related to gene-environment interactions</td>
<td></td>
</tr>
<tr>
<td>Options for genetic testing</td>
<td></td>
</tr>
<tr>
<td>Social and legal issues related to genetic testing (e.g., insurance discrimination)</td>
<td></td>
</tr>
</tbody>
</table>
19. What do you think are the **biggest barriers** to patient access and/or areas of **unmet need** with respect to medical genetic services in Illinois?

20. In your opinion, what are the **biggest issues** public health care providers in local health departments will face over **the next 5 to 10 years** with respect to the provision of genetic services in Illinois?

21. What could the Illinois Department of Public Health do to better support your efforts with respect to the provision of genetic services?
Please provide any other comments you wish to make regarding the delivery of genetic health care services and/or the role of genetics in Illinois’ public health system:

THANK YOU FOR PARTICIPATING IN THIS SURVEY!

Please mail (in enclosed SASE) or fax completed survey to:

Marianne Brennan  
Project Coordinator  
Illinois Regional Health Workforce Center  
University of Illinois, Chicago  
1747 W. Roosevelt Rd., M/C 275  
Chicago, IL 60608  
FAX: (312) 996-0065  
Phone: (312)996-6864  
Email: mbrenn@uic.edu
Appendix 3: Genetic Services Provider Survey  
**ILLINOIS STATEWIDE GENETICS NEEDS ASSESSMENT**  
Survey for Clinical Genetics Service Providers

Please fill in the blank or circle where appropriate.

THANK YOU for your participation!

### I. Provider Practice

1. What is the zip code at your **primary** work location (i.e., where you currently work the most hours)?

2. What is your professional degree(s)?
   - **Select all that apply and indicate year received.**
   - O MD
   - O DO
   - O PhD
   - O MS or MA
   - O Other, specify: 

3. What is your current specialty practice area? **Select all that apply.**
   - O Clinical Biochemical Geneticist
   - O Clinical Molecular Geneticist
   - O Clinical Cytogeneticist
   - O MD or DO Clinical Geneticist
   - O Cytogenetic Technologist
   - O Molecular Technologist
   - O Genetic Counselor
   - O Genetic Nurse
   - O PhD Medical Geneticist
   - O Other, specify: 

4. Do you have any other specialty training? **Select all that apply.**
   - O Cardiology
   - O Endocrinology
   - O Epidemiology
   - O Hematology
   - O Internal Medicine
   - O Neurology
   - O Oncology
   - O Obsterics/Gynecology
   - O Pathology
   - O Pediatrics
   - O Pulmonary
   - O Surgery
   - O Other, specify: 
   - O None

5. How many years have you worked (if retired did you work) in the genetics profession?
   - O I do not work in genetics
   - O Less than 3 years
   - O 3-5 years
   - O 6-10 years
   - O 11-15 years
   - O 16-20 years
   - O 21-25 years
   - O More than 25 years

6. What best describes your **primary** workplace setting (i.e., where you currently work the most hours)? **Select only one response.**
   - **O Academic medical center/university**
   - O Managed care organization/HMO
   - O Medical practice—single specialty
   - O Medical practice—multiple specialties
   - O Commercial laboratory
   - O Pharmaceutical/biotech company
   - O Government agency (non-military)
   - O Consulting (group or self-employed)
   - O Hospital
   - O U.S. military
   - O Other, specify: 

---

Illinois Genetic Services Needs Assessment  
January 2006
7. What is your current rank/title/position? _______________________________________

8. How many hours do you work in an average week? __________________________

9. Please estimate the percentage of time that you spend in the following activities in an average workweek:

<table>
<thead>
<tr>
<th>Percentage Range</th>
<th>Clinical patient care</th>
<th>Clinical laboratory work</th>
<th>Clinical research</th>
<th>Other research</th>
<th>Writing for scientific publications</th>
<th>Teaching/Education</th>
<th>Administration</th>
<th>Other</th>
</tr>
</thead>
<tbody>
<tr>
<td>0%</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>1-10%</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>11-20%</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>21-30%</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>31-40%</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>41-60%</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>61-80%</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>81-99%</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>100%</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
</tbody>
</table>

10. If you spend any time teaching, please estimate how many lectures/presentations you have given to each type of audience over the past 12 months:

<table>
<thead>
<tr>
<th>Audience</th>
<th>Number</th>
</tr>
</thead>
<tbody>
<tr>
<td>Educators (teachers, professors)</td>
<td></td>
</tr>
<tr>
<td>General public</td>
<td></td>
</tr>
<tr>
<td>Medical students/residents/fellows</td>
<td></td>
</tr>
<tr>
<td>Other health professionals</td>
<td></td>
</tr>
<tr>
<td>Support groups/disease organizations</td>
<td></td>
</tr>
<tr>
<td>Students (college undergraduate/graduate)</td>
<td></td>
</tr>
<tr>
<td>Other</td>
<td></td>
</tr>
<tr>
<td>I do not teach</td>
<td></td>
</tr>
</tbody>
</table>
II. Genetics Patient Care/Clinical Practice

11. Please estimate the total number of genetics patient visits you have had in the past 12 months that are:

New outpatients____________________
Return outpatients (seen again within one year of initial visit) ______________
Inpatient consults__________________

12. Of the total number of patients you have seen in the past 12 months, please estimate the percentage with genetic disorders belonging to the following categories:

<table>
<thead>
<tr>
<th>Disorder</th>
<th>0%</th>
<th>1-10%</th>
<th>11-20%</th>
<th>21-30%</th>
<th>31-40%</th>
<th>41-60%</th>
<th>61-80%</th>
<th>81-99%</th>
<th>100%</th>
</tr>
</thead>
<tbody>
<tr>
<td>Dysmorphology/syndromes/birth defects</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Developmental delay/mental retardation</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Metabolic conditions</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Reproductive/prenatal issues (e.g. recurrent miscarriages, advanced maternal age, abnormal serum or ultrasound screening, family history, teratogens)</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Cancer</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Common/complex disorders (e.g. diabetes, asthma, psychiatric disorders)</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Adult-onset single gene disorders (e.g., Huntington’s Disease)</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Other, specify_______________________________</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
</tbody>
</table>

CONTINUE with question 11
13. Approximately what percentage of the genetics patients you have seen over the past 12 months represent:

<table>
<thead>
<tr>
<th>Category</th>
<th>0%</th>
<th>1-10%</th>
<th>11-20%</th>
<th>21-30%</th>
<th>31-40%</th>
<th>41-60%</th>
<th>61-80%</th>
<th>81-99%</th>
<th>100%</th>
</tr>
</thead>
<tbody>
<tr>
<td>Newborns and Infants (less than one year of age)</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Children and Adolescents (not pregnant)</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Prenatal/Reproductive Issues</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Carrier Screening</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Adults (not pregnant)</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
</tbody>
</table>

14. Approximately what percentage of patients referred to you over the past 12 months were referred by:

<table>
<thead>
<tr>
<th>Category</th>
<th>0%</th>
<th>1-10%</th>
<th>11-20%</th>
<th>21-30%</th>
<th>31-40%</th>
<th>41-60%</th>
<th>61-80%</th>
<th>81-99%</th>
<th>100%</th>
</tr>
</thead>
<tbody>
<tr>
<td>MD/DO geneticists</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Generalist physicians (pediatricians, internists, family practitioners)</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Specialist physicians (OBGYN, oncology, neurology)</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Genetic counselors</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Patient (self-referral or family referral)</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Genetic testing laboratories or programs</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>IDPH Genetics and Newborn Screening (NBS follow up)</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Local health departments</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Other, specify:</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Unknown</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
</tbody>
</table>

15. Estimate the percentage of your genetics patients in the following racial groups:

<table>
<thead>
<tr>
<th>Category</th>
<th>0%</th>
<th>1-10%</th>
<th>11-20%</th>
<th>21-30%</th>
<th>31-40%</th>
<th>41-60%</th>
<th>61-80%</th>
<th>81-99%</th>
<th>100%</th>
</tr>
</thead>
<tbody>
<tr>
<td>American Indian or Alaska Native</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Arab or Middle Eastern American</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Asian/Southeast Asian or Pacific Islander</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Black or African American</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>White or Caucasian</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Multiracial (parent from more than one of the above racial or ethnic groups)</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Other</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Unknown</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
</tbody>
</table>
16. Estimate the percentage of your genetics patients in the following ethnic categories:

<table>
<thead>
<tr>
<th></th>
<th>0%</th>
<th>1-10%</th>
<th>11-20%</th>
<th>21-30%</th>
<th>31-40%</th>
<th>41-60%</th>
<th>61-80%</th>
<th>81-99%</th>
<th>100%</th>
</tr>
</thead>
<tbody>
<tr>
<td>Of Hispanic Origin</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Not of Hispanic Origin</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
</tbody>
</table>

17. Estimate the percentage of your genetics patients in each of the following categories of health insurance:

<table>
<thead>
<tr>
<th></th>
<th>0%</th>
<th>1-10%</th>
<th>11-20%</th>
<th>21-30%</th>
<th>31-40%</th>
<th>41-60%</th>
<th>61-80%</th>
<th>81-99%</th>
<th>100%</th>
</tr>
</thead>
<tbody>
<tr>
<td>Traditional indemnity/private insurance</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Managed care (HMO,PPO,IPA)</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Medicaid</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Medicare</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Military</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Uninsured</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Unknown insurance type</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>O Not Applicable</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
</tbody>
</table>

18. What is the typical wait time for an appointment (non-emergency) to see you?  
Existing Patient: ________ days  
New Patient: ________ days

19. Approximately how many missed appointments (no shows) do you experience each week?  
_________________________ # of missed appointments  
_________________________ % of all appointments that are missed

20. How many full-time equivalent staff members does your primary clinical genetics site have?

# FTE  
_______ MD/DO Geneticists (board certified or eligible)  
_______ PhD Geneticists (board certified or eligible)  
_______ Genetic counselors  
_______ Laboratory assistants/support staff  
_______ Technologists/technicians  
_______ Other, specify
21. What is your assessment of the adequacy of staffing at your primary clinical genetics site?

<table>
<thead>
<tr>
<th></th>
<th>Too Few</th>
<th>Right Number</th>
<th>Too Many</th>
<th>Don’t Know</th>
</tr>
</thead>
<tbody>
<tr>
<td>MD/DO Geneticists</td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>PhD Geneticists</td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Genetic Counselors</td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Laboratory assistants/support staff</td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Technologists/technicians</td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Other professionals</td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
</tbody>
</table>

22. Without making substantial changes to your current practice patterns, which response best describes your current genetics practice?

- O I cannot accept any more genetics patients, my genetics practice is full.
- O I can accept some additional genetics patients, my genetics practice is nearly full.
- O I can accept many additional genetics patients, my genetics practice is far from full.

23. Estimate the number of **genetic tests you have recommended or ordered** over the past 12 months.

<table>
<thead>
<tr>
<th># tests</th>
<th></th>
<th></th>
<th></th>
<th></th>
</tr>
</thead>
<tbody>
<tr>
<td>Clinical Biochemical Genetic Tests (excluding AFP and multiple marker)</td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Clinical Molecular Genetic Tests</td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Clinical Cytogenetic Tests</td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Prenatal Screening Samples (AFP, Multiple Marker)</td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Newborn Screening Samples</td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Other Genetic Tests, specify</td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>O Not Applicable</td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
</tbody>
</table>

24. With how many of your patients for whom you recommend/order genetic **susceptibility testing** (for conditions such as familial cancers) or **presymptomatic testing** (for conditions such as Huntington disease) do you discuss:

<table>
<thead>
<tr>
<th></th>
<th>None</th>
<th>Very Few</th>
<th>Several</th>
<th>Many</th>
<th>All</th>
</tr>
</thead>
<tbody>
<tr>
<td>Confidentiality of test results</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Risks of employment discrimination</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Risks of insurance discrimination</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>The potential for inconclusive tests</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Psychological implications of test results</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Testing fees and insurance coverage</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Technical accuracy of test</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>The right to choose not to receive the test</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>O I do not recommend/order genetic susceptibility or presymptomatic genetic tests for my patients</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
</tbody>
</table>
25. Have you ever used any **telemedicine techniques** to provide genetic diagnosis and/or counseling?

O Yes, I believe this is an effective method of service delivery  
O Yes, I think this method of service delivery is somewhat effective  
O Yes, but I do not think this is an effective method of service delivery  
O No, I have not used but think this might be an effective method of service delivery  
O No, I have not used but think this would not be an effective way to deliver genetic services  
O No, but I do provide diagnostic and/or counseling services by telephone.

26. Approximately how many **total outreach clinic sessions** do you travel to yearly?  
*Do not include locations within your primary patient care setting.*

______________________ number per year

O I do not provide outreach services

27. What best describes the **settings** in which you provide outreach services? **Select all that apply.**

O I do not provide outreach services  
O Academic medical center/university  
O Managed care organization/HMO  
O Medical practice—single specialty  
O Medical practice—multiple specialty  
O Commercial laboratory  
O Pharmaceutical/biotechnology company  
O Local, county, or city health department  
O Community health centers  
O Government agency (non-military)  
O Hospital  
O U.S. military  
O Other, specify: ____________________

28. Please identify the **city or county(ies)** in which you provide outreach services:

________________________________________________________________________

III. **Clinical Genetics Laboratory Services**

O I work in a clinical genetics laboratory setting or provide clinical genetics laboratory services  
O I do not work in a clinical genetics laboratory setting or provide clinical genetics laboratory services

**CONTINUE with question 29**  
**SKIP to question 38**
29. In how many clinical genetics laboratories do you work?
   O 1 O 2 O 3 O More than 3

30. Indicate the percentage of clinical laboratory time you spend in each of the following areas:

<table>
<thead>
<tr>
<th>Area</th>
<th>0%</th>
<th>1-10%</th>
<th>11-20%</th>
<th>21-30%</th>
<th>31-40%</th>
<th>41-60%</th>
<th>61-80%</th>
<th>81-90%</th>
<th>100%</th>
</tr>
</thead>
<tbody>
<tr>
<td>Clinical Biochemical Genetics</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Clinical Molecular Genetics</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Clinical Cytogenetics</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Prenatal Screening (AFP, Multiple Marker)</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Newborn Screening</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Non-Genetics Laboratories</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
</tbody>
</table>

31. For your clinical laboratory activities, indicate the percentage of time you spend in each of the following:

<table>
<thead>
<tr>
<th>Activity</th>
<th>0%</th>
<th>1-10%</th>
<th>11-20%</th>
<th>21-30%</th>
<th>31-40%</th>
<th>41-60%</th>
<th>61-80%</th>
<th>81-90%</th>
<th>100%</th>
</tr>
</thead>
<tbody>
<tr>
<td>Test-related activities (benchwork; assay development; assay QC)</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Results interpretation/reports (review/approve assays, reports, etc.)</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Case research/management, follow-up</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Communication with genetic professionals</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Communication with other (non-genetics) healthcare professionals</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Communication with patients</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Other, specify:</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
</tbody>
</table>

32. Estimate the number of tests performed or samples analyzed in the past 12 months at your primary clinical genetics laboratory (i.e., the laboratory in which you work the most hours). Use only those processed to completion by your laboratory. Complete all that apply.

<table>
<thead>
<tr>
<th># tests</th>
</tr>
</thead>
<tbody>
<tr>
<td>____ Clinical Biochemical Genetic Tests (excluding AFP and multiple marker)</td>
</tr>
<tr>
<td>____ Clinical Molecular Genetic Tests</td>
</tr>
<tr>
<td>____ Clinical Cytogenetic Tests</td>
</tr>
<tr>
<td>____ Prenatal Screening Samples (AFP, Multiple Marker)</td>
</tr>
<tr>
<td>____ Newborn Screening Samples</td>
</tr>
<tr>
<td>____ Other Genetic Tests</td>
</tr>
<tr>
<td>____ Non-Genetic Tests</td>
</tr>
</tbody>
</table>
33. Of the number of tests analyzed in your primary clinical genetics laboratory (see #30), what percentage were from/for:

<table>
<thead>
<tr>
<th>Category</th>
<th>0%</th>
<th>1-10%</th>
<th>11-20%</th>
<th>21-30%</th>
<th>31-40%</th>
<th>41-60%</th>
<th>61-80%</th>
<th>81-99%</th>
<th>100%</th>
</tr>
</thead>
<tbody>
<tr>
<td>Newborns and Infants (less than one year of age)</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Children and Adolescents (not pregnant)</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Prenatal/Reproductive genetics patients</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Carrier Screening</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Adults (not pregnant)</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
</tbody>
</table>

34. Over the past 12 months, approximately what percentage of samples submitted to your primary clinical genetics laboratory were submitted by:

<table>
<thead>
<tr>
<th>Party</th>
<th>0%</th>
<th>1-10%</th>
<th>11-20%</th>
<th>21-30%</th>
<th>31-40%</th>
<th>41-60%</th>
<th>61-80%</th>
<th>81-99%</th>
<th>100%</th>
</tr>
</thead>
<tbody>
<tr>
<td>MD/DO geneticists</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Generalist physicians (pediatricians, internists, family practitioners)</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Specialist physicians (OBGYN, oncology, neurology)</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Genetic counselors</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Patient (self-referral or family referral)</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Other genetic testing laboratories or programs</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>IDPH Genetics and Newborn Screening</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Local health departments</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Other</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Unknown</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
</tbody>
</table>

35. Approximately how many specimens has your primary clinical genetics laboratory sent to other labs over the past 12 months?

_________ # to in-state lab
_________ # to out of state labs

36. How many full-time equivalent staff members does your primary clinical genetics lab have?

<table>
<thead>
<tr>
<th># FTE</th>
<th>Description</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td>MD/DO Geneticists (board certified or eligible)</td>
</tr>
<tr>
<td></td>
<td>PhD Geneticists (board certified or eligible)</td>
</tr>
</tbody>
</table>
37. What is your assessment of the adequacy of staffing at your primary clinical genetics laboratory?

<table>
<thead>
<tr>
<th></th>
<th>Too Few</th>
<th>Right Number</th>
<th>Too Many</th>
<th>Don’t Know</th>
</tr>
</thead>
<tbody>
<tr>
<td>MD/DO Geneticists</td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>PhD Geneticists</td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Genetic Counselors</td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Laboratory assistants/support staff</td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Technologists/technicians</td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Other professionals</td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
</tbody>
</table>

IV. Perspective

**All respondents** should reply to the remainder of the survey

38. How familiar are you with Illinois’ Genetics and Newborn Screening Program?
   
   - O Very Familiar
   - O Familiar
   - O Somewhat Familiar
   - O Not Familiar

39. What do you think are the **biggest barriers** to patient access and/or **areas of unmet need** with respect to medical genetic services in Illinois?

40. In your opinion, what are the **biggest issues** genetics providers in your field will face over the **next 5 to 10 years** with respect to the provision of genetic services in Illinois?

41. What could the Illinois Department of Public Health do to better support you as a provider of genetic services?
V. Provider Background

42. What is your gender? O Female O Male

43. What is your year of birth? 19_____

44. What race do you identify with?
   O American Indian or Alaska Native
   O Asian
   O Black or African American
   O Native Hawaiian or Other Pacific Islander
   O White
   O Multiracial (parent from more than one of the above racial or ethnic groups)
   O Other:___________________

45. What ethnicity do you identify with? O Hispanic origin O Not of Hispanic origin

   Please provide any other comments you wish to make regarding the delivery of genetic health care services in Illinois:

THANK YOU FOR PARTICIPATING IN THIS SURVEY!

Please mail (in enclosed SASE) or fax completed survey to:

Marianne Brennan
Project Coordinator
Illinois Regional Health Workforce Center
University of Illinois, Chicago
1747 W. Roosevelt Rd., Room 558, M/C 275
Chicago, Illinois 60608

   FAX: (312) 996-0065
   Phone: (312) 996-6864
   Email: mbrenn@uic.edu
Appendix 4: Interview Guides

Illinois Statewide Genetic Needs Assessment
Interview Guide for Genetic Professionals

*Database Codes:
(primary position, primary work setting)

*Pre-interview Data (to be written by interviewer):

<table>
<thead>
<tr>
<th>*A. Interview Set-Up</th>
</tr>
</thead>
<tbody>
<tr>
<td>*A1. Individual Interviewed (name):</td>
</tr>
<tr>
<td>*A2. Date of interview:</td>
</tr>
<tr>
<td>*A3. Interviewed by:</td>
</tr>
<tr>
<td>Time interview began:</td>
</tr>
<tr>
<td>*A4. Duration of interview:</td>
</tr>
</tbody>
</table>

<table>
<thead>
<tr>
<th>*B. Genetic Services-Description</th>
</tr>
</thead>
<tbody>
<tr>
<td>*B1. Please describe your training and current practice or position.</td>
</tr>
<tr>
<td>*B2. What is your/your organization’s involvement with genetic services in Illinois?</td>
</tr>
<tr>
<td>*B3. Please describe the process by which a typical patient receives services from you. For example, how does the patient get to you? What steps are involved in the services provided by you and others? Do you typically interact with or consult with other medical specialists or health care providers? What kinds of issues do you discuss with a patient? What types of referrals might you make?</td>
</tr>
</tbody>
</table>
| *B4. Could you describe the process of recommending/ordering genetic tests for patients as you experience it? (Interviewer prompts: What do you consider before recommending a test? What do you discuss with the patient? What are commonly expressed patient concerns about testing and how do you respond to these
concerns? How do you decide where to send the tests? What labs do you use?)

*B5. There has been a lot of attention given to the role of primary care providers in the provision of genetic services. In your experience, are primary care providers effectively incorporating genetic information into their practices? How can the roles of primary care physicians and genetic providers better complement each other?

*C. Themes and Critical Issues
In this part of the interview I will be asking you about major challenges and factors that influence your provision of genetic services.

*C1. What do you see as the major challenge(s) to your provision of genetic services?

*C2. In your opinion, what are the biggest issues facing Illinois in the next 5 years, regarding the provision of genetic services?

*C3. How do you see the field of genetics changing over the next five to ten years?

*C4. What do you believe are the greatest barriers to patient access to genetic services in Illinois?

*C5. What genetics services or resources do you think are in greatest need in Illinois?

*C6. What do you think the state department of public health should be doing with respect to the genetic service system in Illinois? What do you see as IDPH’s role in this context? What do you think are the priority issues?

*C7. Any other comments?

Thank you for taking time out of your busy schedule to participate in this study.
Thank you for agreeing to participate in this interview. My name is _____________ and I am a ____________ and interviewer on the research team.

Before we begin, I would like to review some information with you about the interview process. During the interview, I will be asking you a number of questions about genetic services and public health. Our study uses a broad definition of genetic services that includes any activity, including genetic testing, to evaluate, counsel, manage, or plan for an individual or family with a condition that may be inherited or have a genetic basis. In general, this interview takes about 30 minutes.

I would also like to review that you were sent an informed consent form to complete acknowledging your agreement to participate in the telephone interview. You have decided YES/NO. Do you consent to my making an audiotape of our interview, which will only be used for research purposes to allow us to review and clarify any responses? YES/NO. If any of the questions make you uncomfortable, please let me know; you will not be obligated to answer them. Do you have any questions before we begin? YES/NO.

*A. Interview Set-Up
*A1. Individual Interviewed (name):
*A2. Date of interview:
*A3. Interviewed by:
Time interview began:      Time Ended:
*A4. Duration of interview:

B. Genetic Services Description

*B1. Please describe your training and current practice or position.

*B2. What is your/your health department’s current involvement with genetic services in Illinois?

*B3. Could you describe the process by which a typical client receives genetic services from you/your health department. For example, how does the client get to you/your health department? What steps are involved in the genetic/genetics related services provided by you/your health department? Who at your health department is typically involved with providing these services? Do you typically interact with or consult with other medical specialists or health care providers when providing these services? What kinds of issues do you discuss with clients? What types of referrals do you typically make?

*B4. How well do you think IDPH’s system for providing genetic services through local health departments is working? What works? What doesn’t work? What changes would you like to see made to this system?
C. Themes and Critical Issues
In this part of the interview I will be asking you about major challenges and factors that influence your provision of genetic services.

C1. What are some of the major challenges you/your health department face with respect to the provision of genetic services?

C2. How do you think genetics will affect your health department’s practices/programs over the next 5-10 years?

C3. In your opinion, what are the biggest barriers to access with respect to genetic services in Illinois?

C4. What genetics services or resources do you think are in greatest need in Illinois?

C5. What do you think the state department of public health should be doing with respect to the genetic service system in Illinois? What do you see as IDPH’s role? What do you think are the priority issues?

C6. Any other comments?

Thank you for taking time out of your busy schedule to participate in this study.
Illinois Statewide Genetic Needs Assessment
Interview Guide for Health Care Providers

*Database Codes:
(Primary position, primary work setting)

*Pre-interview Data (to be written by interviewer):

Thank you for agreeing to participate in this interview. My name is _____________ and I am a ____________ and interviewer on the research team.

Before we begin, I would like to review some information with you about the interview process. During the interview, I will be asking you a number of questions about health care and genetic services. Our study uses a broad definition of genetic services that includes any activity, including genetic testing, to evaluate, counsel, manage, or plan for an individual or family with a condition that may be inherited or have a genetic basis. In general, this interview takes about 30 minutes.

I would also like to review that you were sent an informed consent form to complete acknowledging your agreement to participate in the telephone interview. You have decided YES/NO. Do you consent to my making an audiotape of our interview, which will only be used for research purposes to allow us to review and clarify any responses? YES/NO. If any of the questions make you uncomfortable, please let me know; you will not be obligated to answer them. Do you have any questions before we begin? YES/NO.

*A. Interview Set-Up
*A1. Individual Interviewed (name):
*A2. Date of interview:
*A3. Interviewed by:
  Time interview began:      Time Ended:
*A4. Duration of interview:

*B. Genetic Services and Health Care-Description

*B1. Please describe your training and current practice or position.

*B2. How has the expansion of medical genetics affected your practice over the past 5-10 years? What impact do you anticipate it will have in the next 5 years on your medical practice?

*B3. As a ____________ (Select most appropriate: physician, obstetrician/gynecologist, advanced practice nurse, pediatrician, internist, cardiologist, neurologist, oncologist, hematologist, psychiatrist/psychologist), what do you see as your role in the genetic service system in Illinois?

*B4. Please describe the process by which a typical patient receives genetic services from you? For example, how does the issue of genetics typically come up? What steps are involved in the services provided by you? Do you typically interact with or consult with other medical specialists or health care providers? What kinds of things do you discuss with a patient?

*B5. Where and to whom might you refer a patient for genetic counseling or diagnostics?
**B6.** How many genetic tests do you estimate you have recommended or ordered over the past 6 months?

**B7.** Please describe the process of recommending/ordering genetic tests for patients as you experience it? (Interviewer prompts: What do you consider before recommending a test? What do you discuss with the patient? What are commonly expressed patient concerns about testing? How do you respond to these concerns? How do you decide where to send the tests? What labs do you use?)

**C. Themes and Critical Issues**

In this part of the interview I will be asking you about major challenges and factors that influence your provision of genetic services.

**C1.** What are some of the major challenges you face with respect to the provision of genetic services?

**C2.** In your opinion, what are the biggest issues facing Illinois in the next 5 years with respect to the provision of genetic services?

**C3.** What do you believe are the biggest barriers to access with respect to genetic services in Illinois?

**C4.** What genetics services or resources do you think are in greatest need in Illinois?

**C5.** What do you think the state department of public health should be doing with respect to the genetic service system in Illinois? What do you see as IDPH’s role? What do you think are the priority issues?

**C6.** Any other comments?

Thank you for taking time out of your busy schedule to participate in this study.
Thank you for agreeing to participate in this interview. My name is ___________ and I am a ___________ and interviewer on the research team.

Before we begin, I would like to review some information with you about the interview process. During the interview, I will be asking you a number of questions about genetic services and public health. Our study uses a broad definition of genetic services that includes any activity, including genetic testing, to evaluate, counsel, manage, or plan for an individual or family with a condition that may be inherited or have a genetic basis. In general, this interview takes about 30 minutes.

I would also like to review that you were sent an informed consent form to complete acknowledging your agreement to participate in the telephone interview. You have decided YES/NO. Do you consent to my making an audiotape of our interview, which will only be used for research purposes to allow us to review and clarify any responses? YES/NO. If any of the questions make you uncomfortable, please let me know; you will not be obligated to answer them. Do you have any questions before we begin? YES/NO.

*A. Interview Set-Up
* A1. Individual Interviewed (name):
* A2. Date of interview:
* A3. Interviewed by:
Time interview began: Time Ended:
* A4. Duration of interview:

*B. Programmatic Impact
* B1. Please describe your training and current practice or position.

* B2. How would you describe your knowledge/experience in the field of genetics or genomics?
   Minimal to None (e.g., don’t feel I have a good understanding of concepts or issues)
   Limited (e.g., understand many basic concepts, but would have difficulty applying to public health practice)
   Average (e.g., understand basic and some complex concepts, but might have difficulty applying to public health practice)
   Above Average (e.g., understand basic and many complex concepts, may be able to apply concepts to practice)
   Advanced (e.g., have formal training/understanding of genetics concepts and issues and can apply genetics concepts to practice)

* B3. Are genetic issues currently addressed in your program area? If so, what issues are being addressed and how?

* B4. How do you think genetics information or services might (now or in the near future) benefit your
program and the clients you serve? What opportunities do you see for integration of genetics into your program area?

*B5. What barriers do you see for integration of genetics into your program area?

*C. Themes and Critical Issues

In this part of the interview I will be asking you about major challenges and factors affecting the provision of genetic services in Illinois.

*C1. What do you think are or will be the major genetics or genetics related issues public health will need to address in the next five years?

*C2. What genetics or genetics-related services or resources do you think are in greatest need in Illinois?

*C3. In your opinion, what are the biggest barriers to access with respect to genetic services in Illinois?

*C4. What do you think the state department of public health should be doing with respect to the genetic service system in Illinois? What do you see as IDPH’s role? What do you think are the priority issues?

*C5. Any other comments?

Thank you for taking time out of your busy schedule to participate in this study.
**Illinois Statewide Genetic Needs Assessment**  
**Interview Guide for Advocacy Groups/Consumers**

*Database Codes:*
(*primary position, primary work setting*)

*Pre-interview Data (to be written by interviewer):*

---

Thank you for agreeing to participate in this interview. My name is ______________ and I am a ___________ and interviewer on the research team.

Before we begin, I would like to review some information with you about the interview process. During the interview, I will be asking you a number of questions about genetic services and public health. Our study uses a broad definition of genetic services that includes any activity, including genetic testing, to evaluate, counsel, manage, or plan for an individual or family with a condition that may be inherited or have a genetic basis. In general, this interview takes about 30 minutes.

I would also like to review that you were sent an informed consent form to complete acknowledging your agreement to participate in the telephone interview. You have decided YES/NO. Do you consent to my making an audiotape of our interview, which will only be used for research purposes to allow us to review and clarify any responses? YES/NO. If any of the questions make you uncomfortable, please let me know; you will not be obligated to answer them. Do you have any questions before we begin? YES/NO.

---

**A. Interview Set-Up**

* A1. Individual Interviewed (name):
* A2. Date of interview:
* A3. Interviewed by:
  
  Time interview began:      Time Ended:

* A4. Duration of interview:

**B. Genetic Services Description**

* B1. Please describe your training and current practice or position.

* B2. How would you describe your knowledge/experience in the field of genetics or genomics?

* B3. What is your/your organization’s current involvement with genetic services in Illinois?

* B4. How does your organization interact with IDPH? With other state agencies?

* B5. How do you communicate with your constituency about genetic issues?

* B6. What are some of the key areas of concern for consumers in utilizing genetic services?  
  *Prompts: Do consumers know where to go for information? Do consumers know where to go for genetic services? How do they find these services? Do health care providers know about available genetic and social services? What barriers do they face when attempting to access these services?*
*B7. What do you see as some strategies for addressing these concerns?

*B8. How do you think genetics information or services might (now or in the near future) benefit your organization and your constituents?

*C. Themes and Critical Issues
In this part of the interview I will be asking you about major challenges and factors that influence your provision of genetic services.

*C1. What do you think are or will be the major genetics or genetics related issues public health will need to address in the next five years?

*C2. What genetics or genetics-related services or resources do you think are in greatest need in Illinois?

*C3. In your opinion, what are the biggest barriers to access with respect to genetic services in Illinois?

*C4. What do you think the state department of public health should be doing with respect to the genetic service system in Illinois? What do you see as IDPH’s role? What do you think are the priority issues?

*C5. Any other comments?

Thank you for taking time out of your busy schedule to participate in this study.
Appendix 5: List of Disorders Screened

- Biotinidase Deficiency
- Congenital Adrenal Hyperplasia
- Galactosemia
- Congenital Hypothyroidism
- Sickle Cell Disease and other Hemoglobinopathies

**Amino Acid Disorders**
- Phenylketonuria (PKU)
- Maple syrup urine disease (MSUD)
- Tyrosinemia types I and possibly type II or III (TYRO)
- Homocystinuria (HCU)
- Oxoprolinuria (5OXP)
- Citrullinemia (CIT)
- Argininosuccinic aciduria (ASA)
- Argininemia (ARG)

**Fatty Acid Oxidation Disorders**
- Short chain acyl-CoA dehydrogenase (SCAD)
- Isobutyryl-CoA dehydrogenase deficiency (IBCD)
- Medium/short chain L-3 hydroxyacyl-CoA dehydrogenase deficiency (M/SCHAD)
- Very long chain acyl-CoA dehydrogenase
- Carnitine palmitoyl transferase deficiency type IA and II (CPTIA, CPTII)
- Trifunctional protein deficiency (TFPD)
- Long chain 3-hydroxy acyl-CoA dehydrogenase (LCHAD)
- Glutaric Aciduria type II or multiple acyl-CoA dehydrogenase deficiency (GAI)
- Carnitine/acylcarnitine translocase deficiency (CACT)

**Organic Acid Disorders**
- 3-methylcrotonyl-CoA carboxylase deficiency (3MCC)
- 3-hydroxy-3-methylglutaric-CoA lyase deficiency (3HMG)
- 3-methylglutaconic aciduria (3MGA)
- Multiple carboxylase deficiency (MCD)
- 2-methyl-butryryl-CoA dehydrogenase deficiency (2MBCD)
- Isovaleric acidemia (IVA)
- Methylmalonic acidemia (MMA)
- Malonic aciduria (MA)
- Propionic acidemia (PA)
- Beta-ketothiolase deficiency (BKT)
- Glutaric aciduria type I (GAI)

*Source: IDPH, Division of Laboratories. Manual of Services. February 2005, p. 28-29. Available online at: [http://www.idph.state.il.us/about/laboratories/labman.pdf](http://www.idph.state.il.us/about/laboratories/labman.pdf). Updated by IDPH Genetics and Newborn Screening staff January 11, 2006. For more information on these conditions, see the IDPH Genetics and Newborn Screening Program website at: [http://www.idph.state.il.us/HealthWellness/genetics.htm](http://www.idph.state.il.us/HealthWellness/genetics.htm)*
Appendix 6: List of IDPH Local Health Department Grantees

Effective Date: July 1, 2004-June 30, 2005, *Bold Indicates Genetic Outreach Clinics*

<table>
<thead>
<tr>
<th>Health Department</th>
<th>Area Covered/Genetics Clinic</th>
</tr>
</thead>
<tbody>
<tr>
<td>Bond County Health Department</td>
<td>Bond</td>
</tr>
<tr>
<td>503 S. Prairie St.</td>
<td></td>
</tr>
<tr>
<td>Greenville, IL  62246</td>
<td></td>
</tr>
<tr>
<td>Bureau County Health Department</td>
<td>Bureau, Putnam</td>
</tr>
<tr>
<td>526 Bureau Valley Parkway</td>
<td></td>
</tr>
<tr>
<td>Princeton, IL  61356</td>
<td></td>
</tr>
<tr>
<td>Champaign-Urbana Public Health District</td>
<td>Clark, Coles, Cumberland, Douglas, Ford, Iroquois, Vermilion</td>
</tr>
<tr>
<td>710 N. Neil St.</td>
<td>City of Champaign and Urbana</td>
</tr>
<tr>
<td>Champaign, IL  61820-1488</td>
<td></td>
</tr>
<tr>
<td>Chicago Department of Health</td>
<td>City of Chicago</td>
</tr>
<tr>
<td>DePaul University Center</td>
<td></td>
</tr>
<tr>
<td>333 S. State St., Rm. 200</td>
<td></td>
</tr>
<tr>
<td>Chicago, IL  60604</td>
<td></td>
</tr>
<tr>
<td>Christian County Health Department</td>
<td>Christian</td>
</tr>
<tr>
<td>902 W. Springfield Road</td>
<td></td>
</tr>
<tr>
<td>Taylorville, IL 62568</td>
<td></td>
</tr>
<tr>
<td>Cook County Department of Public Health</td>
<td>Suburban Cook</td>
</tr>
<tr>
<td>1010 Lake St., Suite 300</td>
<td></td>
</tr>
<tr>
<td>Oak Park, IL  60301</td>
<td></td>
</tr>
<tr>
<td>Crawford County Health Department</td>
<td>Crawford</td>
</tr>
<tr>
<td>202 N. Bline Blvd.</td>
<td></td>
</tr>
<tr>
<td>Robinson, IL  62454</td>
<td></td>
</tr>
<tr>
<td>DuPage County Health Department</td>
<td>DuPage</td>
</tr>
<tr>
<td>111 N. County Farm Road</td>
<td></td>
</tr>
<tr>
<td>Wheaton, IL  60187</td>
<td></td>
</tr>
<tr>
<td>East Side Health District</td>
<td>St. Clair (E. St. Louis, Washington Park, Fairmount City, Southwest area of Caseyville, Cahokia, Alorton, Centreville, Sauget, Brooklyn, Lovejoy)</td>
</tr>
<tr>
<td>638 N. 20th St.</td>
<td></td>
</tr>
<tr>
<td>East St. Louis, IL  62205</td>
<td></td>
</tr>
<tr>
<td>Edgar County Health Department</td>
<td>Edgar</td>
</tr>
<tr>
<td>502 Shaw Ave.</td>
<td></td>
</tr>
<tr>
<td>Paris, IL  61944</td>
<td></td>
</tr>
<tr>
<td>Egyptian County Health Department</td>
<td>Gallatin, Saline, White</td>
</tr>
<tr>
<td>Rt. 3, Box 90A, 1412 U.S. 45 North</td>
<td></td>
</tr>
<tr>
<td>Eldorado, IL 62930-9234</td>
<td></td>
</tr>
<tr>
<td>Evanston Health Department</td>
<td>City of Evanston (Cook County)</td>
</tr>
<tr>
<td>Evanston Civic Center</td>
<td></td>
</tr>
<tr>
<td>2100 Ridge Ave.</td>
<td></td>
</tr>
<tr>
<td>Evanston, IL  60201</td>
<td></td>
</tr>
<tr>
<td><strong>Fayette County Health Department</strong>*</td>
<td>Effingham, Fayette, Jasper, Lawrence General Genetic Clinic 2 x yearly – St. Louis University</td>
</tr>
<tr>
<td>---------------------------------------</td>
<td>----------------------------------------------------------------------------------</td>
</tr>
<tr>
<td>P.O. Box 340</td>
<td></td>
</tr>
<tr>
<td>509 W. Edwards St.</td>
<td></td>
</tr>
<tr>
<td>Vandalia, IL  62471</td>
<td></td>
</tr>
<tr>
<td></td>
<td></td>
</tr>
<tr>
<td>Henderson County Health Department</td>
<td>Henderson, Warren</td>
</tr>
<tr>
<td>P.O. Box 220</td>
<td></td>
</tr>
<tr>
<td>Gladstone, IL 61437-0220</td>
<td></td>
</tr>
<tr>
<td></td>
<td></td>
</tr>
<tr>
<td>Henry County Health Department</td>
<td>Henry, Stark</td>
</tr>
<tr>
<td>4424 U.S. Highway 34</td>
<td></td>
</tr>
<tr>
<td>Kewanee, IL  61443</td>
<td></td>
</tr>
<tr>
<td></td>
<td></td>
</tr>
<tr>
<td><strong>Jackson County Health Department</strong>*</td>
<td>Frankin-Williamson, Jackson, Perry General Genetics Clinic every month - St. Louis University</td>
</tr>
<tr>
<td>Route 13 at Country Club Rd.</td>
<td></td>
</tr>
<tr>
<td>P.O. Box 307</td>
<td></td>
</tr>
<tr>
<td>Murphysboro, IL 62966</td>
<td></td>
</tr>
<tr>
<td></td>
<td></td>
</tr>
<tr>
<td><strong>Jefferson County Health Department</strong>*</td>
<td>Jefferson General Genetics Clinic - 6 x yearly – St. Louis University</td>
</tr>
<tr>
<td>#1 Doctors Park Rd., Ste. F</td>
<td></td>
</tr>
<tr>
<td>Mt. Vernon, IL  62864</td>
<td></td>
</tr>
<tr>
<td></td>
<td></td>
</tr>
<tr>
<td>Jersey County Health Department</td>
<td>Calhoun, Greene, Jersey</td>
</tr>
<tr>
<td>1307 State Highway 109</td>
<td></td>
</tr>
<tr>
<td>Jerseyville, IL 62052</td>
<td></td>
</tr>
<tr>
<td></td>
<td></td>
</tr>
<tr>
<td>Kane County Health Department</td>
<td>Kane</td>
</tr>
<tr>
<td>1240 N. Highland Ave., Suite 12</td>
<td></td>
</tr>
<tr>
<td>Aurora, IL  60506</td>
<td></td>
</tr>
<tr>
<td></td>
<td></td>
</tr>
<tr>
<td>Kankakee County Health Department</td>
<td>Kankakee</td>
</tr>
<tr>
<td>2390 W. Station Street</td>
<td></td>
</tr>
<tr>
<td>Kankakee, IL 60901</td>
<td></td>
</tr>
<tr>
<td></td>
<td></td>
</tr>
<tr>
<td>Knox County Health Department</td>
<td>Knox</td>
</tr>
<tr>
<td>1361 West Fremont St.</td>
<td></td>
</tr>
<tr>
<td>Galesburg, IL 61401</td>
<td></td>
</tr>
<tr>
<td></td>
<td></td>
</tr>
<tr>
<td><strong>LaSalle County Health Department</strong>*</td>
<td>LaSalle General Genetics Clinic 4 x yearly – University of IL Medical Center</td>
</tr>
<tr>
<td>717 Etna Rd.</td>
<td></td>
</tr>
<tr>
<td>Ottawa, IL 61350</td>
<td></td>
</tr>
<tr>
<td></td>
<td></td>
</tr>
<tr>
<td><strong>Macon County Health Department</strong></td>
<td>DeWitt, Livingston, Macon, Moultrie, Piatt, Shelby Sickle Cell Clinic 2 x yearly - University of Peoria Medical Center</td>
</tr>
<tr>
<td>1221 E. Condit St.</td>
<td></td>
</tr>
<tr>
<td>Decatur, IL 62521-1405</td>
<td></td>
</tr>
<tr>
<td></td>
<td></td>
</tr>
<tr>
<td><strong>Macoupin County Health Department</strong>*</td>
<td>Macoupin General Genetic Clinic 4 x yearly - SIU Medical Center, Springfield</td>
</tr>
<tr>
<td>805 N. Broad St.</td>
<td></td>
</tr>
<tr>
<td>Carlinville, IL 62626</td>
<td></td>
</tr>
<tr>
<td></td>
<td></td>
</tr>
<tr>
<td><strong>Madison County Health Department</strong>*</td>
<td>Madison General Genetics Clinic 6 x yearly – St. Louis University</td>
</tr>
<tr>
<td>101 E. Edwardsville Road</td>
<td></td>
</tr>
<tr>
<td>Wood River, IL 62095</td>
<td></td>
</tr>
<tr>
<td></td>
<td></td>
</tr>
<tr>
<td>Marion County Health Department</td>
<td>Marion</td>
</tr>
<tr>
<td>600 E. Main St.</td>
<td></td>
</tr>
<tr>
<td>Salem, IL 62881</td>
<td></td>
</tr>
<tr>
<td>Health Department</td>
<td>County</td>
</tr>
<tr>
<td>-------------------------------------------------------</td>
<td>--------------</td>
</tr>
<tr>
<td>McDonough County Health Department 505 E. Jackson St.</td>
<td>McDonough</td>
</tr>
<tr>
<td>Macomb, IL  61455</td>
<td></td>
</tr>
<tr>
<td>McLean County Health Department 200 W. Front St., Rm.</td>
<td>McLean</td>
</tr>
<tr>
<td>304 Bloomington, IL  61701</td>
<td></td>
</tr>
<tr>
<td>Mercer County Health Department 1007 NW 3rd St.</td>
<td>Mercer</td>
</tr>
<tr>
<td>Aledo, IL  61231</td>
<td></td>
</tr>
<tr>
<td>Monroe-Randolph Bi-County Health Department 2515 State</td>
<td>Monroe,</td>
</tr>
<tr>
<td>St. Chester, IL  62233</td>
<td>Randolph</td>
</tr>
<tr>
<td>Montgomery County Health Department 11191 Illinois</td>
<td>Montgomery</td>
</tr>
<tr>
<td>Route 185 Hillsboro, IL  62049-0128</td>
<td>General</td>
</tr>
<tr>
<td>Genetics Clinic 2 x yearly - SIU Medical Center,</td>
<td>Springfield</td>
</tr>
<tr>
<td>Springfield</td>
<td></td>
</tr>
<tr>
<td>Rock Island County Health Department 2112 25th Ave.</td>
<td>Rock Island</td>
</tr>
<tr>
<td>Rock Island, IL  61201</td>
<td></td>
</tr>
<tr>
<td>St. Clair County Health Department #19 Public Square,</td>
<td>St. Clair</td>
</tr>
<tr>
<td>Ste. 150 Belleville, IL  62220</td>
<td>(Caseyville,</td>
</tr>
<tr>
<td>Dupo, Southern Tip of Collinsville and rest of St.</td>
<td></td>
</tr>
<tr>
<td>Clair County except as covered by East Side Health</td>
<td></td>
</tr>
<tr>
<td>District)</td>
<td></td>
</tr>
<tr>
<td>Sangamon County Department of Public Health 2501 N.</td>
<td>Brown, Cass</td>
</tr>
<tr>
<td>Dirksen Parkway Springfield, IL  62702</td>
<td>Logan, Mason,</td>
</tr>
<tr>
<td>Menard, Morgan, Pike, Sangamon, Schuyler, Scott</td>
<td>Pope, Pulaski,</td>
</tr>
<tr>
<td></td>
<td>Union</td>
</tr>
<tr>
<td>Southern Seven Health Department P. O. Box 817</td>
<td>Alexander,</td>
</tr>
<tr>
<td>Roseclair, IL  62982</td>
<td>Hardin,</td>
</tr>
<tr>
<td></td>
<td>Johnson,</td>
</tr>
<tr>
<td></td>
<td>Massac,</td>
</tr>
<tr>
<td></td>
<td>Pope,</td>
</tr>
<tr>
<td></td>
<td>Pulaski,</td>
</tr>
<tr>
<td></td>
<td>Union</td>
</tr>
<tr>
<td>Springfield Department of Public Health* 1415 E.</td>
<td>Springfield</td>
</tr>
<tr>
<td>Jefferson St. Springfield, IL  62703</td>
<td>City</td>
</tr>
<tr>
<td>General Genetics Clinic 4 x yearly - University of</td>
<td></td>
</tr>
<tr>
<td>IL, Chicago Medical Center</td>
<td></td>
</tr>
<tr>
<td>Tazewell County Health Department 21306 Illinois</td>
<td>Tazewell</td>
</tr>
<tr>
<td>Route 9 Tremont, IL  61568-9252</td>
<td></td>
</tr>
<tr>
<td>Will County Health Department* 501 Ella Ave.</td>
<td>Will, Grundy,</td>
</tr>
<tr>
<td>Joliet, IL  60433</td>
<td>Kendall</td>
</tr>
<tr>
<td>Genetics Clinic 6 x yearly – Advocate Christ Hospital</td>
<td></td>
</tr>
<tr>
<td>Winnebago County Health Department 401 Division St.</td>
<td>Boone, Carroll, DeKalb, JoDaviess, Lee, Ogle,</td>
</tr>
<tr>
<td>Rockford, IL  61104</td>
<td>Stephenson,</td>
</tr>
<tr>
<td></td>
<td>Whiteside,</td>
</tr>
<tr>
<td></td>
<td>Winnebago</td>
</tr>
</tbody>
</table>
Appendix 7. IDPH EXPANDED GENETIC SCREENING TOOL

A positive response should be reviewed by a genetic coordinator and does not necessarily indicate a referral to a genetic center. See Rationale for

<table>
<thead>
<tr>
<th>If respondent is different than client, please indicate name and relationship to client.</th>
<th>You</th>
<th>Your Partner</th>
<th>Your Child/Children</th>
<th>Other Family Members</th>
<th>Comments</th>
</tr>
</thead>
<tbody>
<tr>
<td>1. Medical problems since birth or birth defect (cleft lip/palate, spina bifida, heart defects, ambiguous genitalia, etc.)</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>2. A child with any unusual facial appearance or physical features</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>3. Learning problems, developmental delay, mental retardation</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>4. A child with regression of developmental skills</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>5. A child with abnormal growth (height, weight, or head circumference)</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>6. A stillborn child or an early infant death</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>7. A child with a confirmed positive newborn screening test</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>8. Two or more first trimester miscarriages</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>9. You and your partner are blood relatives</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>10. Chromosomal abnormalities (Down syndrome, trisomy 13, Turner Syndrome, etc.)</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>11. Neurological or muscular diseases (muscular dystrophies, Huntington’s disease, etc.)</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>12. Disorders of the blood (sickle cell disease, thalassemia, hemophilia, Factor V Leiden, etc.)</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>13. Vision loss (not glasses) or hearing loss at an early age</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>14. Abnormalities of the bones or skin (brittle bones, bone deformities, unusual birth marks)</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>15. Other genetic diseases (cystic fibrosis, polycystic kidney disease, Tay Sachs disease, newborn screening disorder, i.e., PKU or phenylketonuria)</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
</tbody>
</table>

Client’s Name_________________________________________ County of Residence____________________________________
Race/Ethnicity: Client _______ Partner
Birth Date___________________________ Age_____Sex_____ Agency/Program Name___________________________________
Religion: Jewish     Non-Jewish  ( please circle one)
Date of Assessment_____________________________________ Staff Person Completing Form _____________________________
16. A. Breast, ovarian, colon, skin cancer or a history of colon polyps prior to age 50?
   B. An evaluation and/or diagnosis for any cancer conditions

17. A. A heart attack, stroke or sudden early death (women <60 yrs; men <50 yrs)
   B. An evaluation and/or diagnosis for a heart condition
   C. High blood pressure and/or high cholesterol

18. Diabetes (Type 1, Type 2 or Maturity Onset Diabetes of the Young (MODY)

19. Asthma

20. Arthritis

*Clients with a positive response regarding questions #16 and #17 should only be referred if they are high risk or have a significant concern. Please see rationale for details.

**At this time, clients with a positive response regarding questions *18, #19 and #20, do not need a genetic referral but may be offered educational materials. Please see rationale for details.

ADDITIONAL QUESTIONS FOR WOMEN: For any prenatal case, a positive response requires an immediate referral to the genetic coordinator to your local genetic center, except for questions 5 and 6.

Mark an X if any of the following apply to the respondent

1. Are you over the age of 35 years?
2. Do you have diabetes or a history of gestational diabetes?
3. Do you have seizures or being treated for seizures?
4. Are you currently taking prescription drugs?
5. Are you currently taking vitamins containing folic acid?
6. Do you have concerns about exposures to cigarette smoking?
7. Do you have concerns about alcohol consumption or drug use?
8. Do you have concerns about exposures to harmful substances in the workplace?
9. If you are pregnant, have you had chicken pox, rubella or CMV during this pregnancy?
10. If you are pregnant, have you had an abnormal 1st trimester or maternal serum multiple marker screen?
11. If you are pregnant, have you had an abnormal finding on an ultrasound examination?
12. Have you had abnormal results from a CVS or an amniocentesis?

If pregnant: Name of OB_______________________ LMP___________ EDC___________ Gravida______ Para______

Does the client wish to receive further information about any answers that triggered a yes response? Yes or No
Would they be interested in seeing a Genetic Counselor? Yes or No
Can client be contacted by Genetic Case Manager? Yes or No. Are they already being followed by a Geneticist/Genetic Counselor? Yes or No. Condition or Diagnosis: _______________________
Client prefers to be contacted by Phone or Mail. Phone Number: ______________________ Address: ______________________

**Disposition by Genetic Coordinator**

<table>
<thead>
<tr>
<th>No referral indicated ______</th>
<th>Referral Made: Date <strong>/</strong>/____</th>
<th>Appointment Kept____</th>
<th>Missed____</th>
<th>Referred to: ____________________________</th>
</tr>
</thead>
<tbody>
<tr>
<td>REFERRAL INDICATED _____</td>
<td>Follow-Up Attempts: Indicate: Phone (P), Mail (M) Home Visit (HV)</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>REFUSED: Date: <strong>/</strong>/____</td>
<td>1&lt;sup&gt;st&lt;/sup&gt; Attempt: Date: <strong>/</strong>/____</td>
<td>2&lt;sup&gt;nd&lt;/sup&gt; Attempt: Date: <strong>/</strong>/____</td>
<td>3&lt;sup&gt;rd&lt;/sup&gt; Attempt: Date: <strong>/</strong>/____</td>
<td></td>
</tr>
<tr>
<td>If being followed by geneticist, indicate provider:</td>
<td>Additional Comments/Informational handouts provided:</td>
<td></td>
<td></td>
<td></td>
</tr>
</tbody>
</table>

IL#482-0962 Printed by Authority of the State of Illinois P.O.
Appendix 8. Genetics and Metabolic Diseases Advisory Committee

Carol Booth, M.D.
Section of Genetics
Lutheran General Perinatal Center
1875 Dempster, Suite 340
Park Ridge, Illinois  60068

Barbara K. Burton, M.D.
Division of Genetics (MC 59)
Children’s Memorial Hospital
2300 Children’s Plaza
Chicago, Illinois  60614

James Critchfield
1309 Hampton Place
Palatine, Illinois  60067

Shelly Cummings
801 S. Plymouth Court #618
Chicago, Illinois  60605

Rich Dineen, M.S.
Division of Genetics (MC 857)
The University of Illinois at Chicago
820 S. Wood St.
Chicago, Illinois  60612

George Hoganson, M.D.
Genetics and Metabolism, MC 856
University of Illinois Hospital
840 S. Wood St.
Chicago, Illinois  60612

Robert Klutts
Southern Illinois Healthcare Found., Inc.
8080 State St.
East St. Louis, Illinois  62203

Anne C. Kozek
Division of Genetics (MC 857)
The University of Illinois at Chicago
820 S. Wood St.
Chicago, Illinois  60612

Rodney A. Lorenz, M.D.
Department of Pediatrics
530 NE Glen Oak Ave.
Peoria, Illinois  61637

Candace Madison
6025 W. Beechwood Road
Matteson, Illinois  60443

Debra E. Weese-Mayer, M.D.
Pediactric Respiratory Medicine
Rush Children’s Hospital
1653 W. Congress Parkway
Chicago, Illinois  60612

Sunetra Reddy
University of Chicago Hospitals
Laboratory Referral Service
5841 S. Maryland Ave., RM. TW005
Chicago, Illinois  60637

Barry Rich, M.D.
Section of Pediatrics Endocrinology
University of Chicago
East 65th St. at Lake Michigan
Chicago, Illinois  60649

Kay L. Saving, M.D.
530 N.E. Glen Oak Ave.
Peoria, Illinois  61637

Debby Smith, R.N.
Fayette County Health Department
P.O. Box 340, 509 W. Edwards
Vandalia, Illinois  62471

Hazel Vespa, L.C.S.W.
Social Services/Family Support Serv., #130
Children’s Memorial Hospital
2300 Children’s Plaza
Chicago, Illinois  60614

Cari Vonderhaar, R.N.
Bond County Health Department
503 S. Prairie St.
Greenville, Illinois  62246

Christine Weaver, M.D., Ph.D.
Department of Pediatrics
Carle Clinic
601 W. University
Urbana, Illinois  61801
Ex Officio Members:

Mark Schmidt, Chief  
IDPH, Health Assessment & Screening  
Ridgely Bldg., 1st Fl., 500 E. Monroe  
Springfield, Illinois 62701

Judy Miller, M.S.  
Carle Clinic, Medical Genetics/AM S5  
602 W. University  
Urbana, Illinois 61801

Sharon Green, Deputy Director  
IDPH, Office of Women’s Health  
100 W. Randolph, Suite 6-600  
Chicago, Illinois 60601

Margaret Hastings, Ph.D.  
366 Sterling Road  
Kenilworth, Illinois 60043

David C. Jinks, Ph.D., Chief  
IDPH, Newborn Screening Lab  
2121 W. Taylor St., P.O. Box 12279  
Chicago, Illinois 60612

Tom Johnson  
Illinois Department of Public Health  
825 N. Rutledge St.  
Springfield, Illinois 62794-9635

Lew Lampiris, D.D.S., M.P.H.  
Assistant Deputy Director  
IDPH, Office of Health Promotion  
100 W. Randolph, Suite 6-600  
Chicago, Illinois 60601

Susan Marantz, M.D., Director  
IDPH, Bureau of Medical Programs  
100 W. Randolph, Suite 6-600  
Chicago, Illinois 60601

David Maserang, Ph.D.  
Laboratory Director, IDPH  
825 N. Rutledge  
Springfield, Illinois 62794

Chandana Nandi, MS, RD, LD  
IDPH, Chronic Disease Prevention  
535 W. Jefferson Street, 2nd Fl.  
Springfield, Illinois 62761

Claudia Nash, M.S.  
IDPH, Genetics Newborn Screening  
Ridgely Bldg., 1st Fl.  
500 E. Monroe  
Springfield, Illinois 62701

Charles Onufer, M.D., Director  
Division of Specialized Care for Children  
2815 W. Washington, Suite 300  
P.O. Box 19481  
Springfield, Illinois 62794-9481

Eugene Pergament, M.D., Ph.D.  
680 N. Lake Shore Drive, Suite 1230  
Chicago, IL 60611

Mike Petros  
IDPH, State of Illinois Neonatal Screening  
2121 W. Taylor, P.O. Box 12279  
Chicago, IL 60612

Marilyn Thomas, J.D.  
IDPH, Division of Legal Services  
535 W. Jefferson St., 5th Floor  
Springfield, Illinois 62761

W. Patrick Zeller, M.D.  
Clinical Professor Pediatrics  
Loyola University of Chicago  
120 Spalding, Suite #401  
Naperville, Illinois 60540

Lisa Dye, M.Ed.  
March of Dimes  
Greater Illinois Chapter  
111 W. Jackson Blvd., 22nd Fl.  
Chicago, Illinois 60604

Joseph DeSimone, Ph.D.  
UIC Section of Hematology/Oncology  
Department of Medicine  
840 S. Wood St.  
Chicago, Illinois 60612-7323

Valerie Beckley  
Sickle Cell Disease Association of Illinois  
200 N. Michigan Ave., Suite 605  
Chicago, Illinois 60601-5908

ADVCOMLIST04  03/09/04
Appendix 9: Catalogue of On-going Medical Genetics Projects in Illinois, by Institution

Chicago Center for Jewish Genetic Disorders

Education and Screening Program  
Funder: Jewish Federation of Metropolitan Chicago  
Director: Karen Litwack  
This project provides education about and screening for genetic disorders affecting Ashkenazi Jews to target populations 18-35 years old.

Loyola University

Genetics of Hypertension in Blacks  
Funder: National Heart, Lung, and Blood Institute  
PI: Richard Stanley Cooper  
This study conducts linkage analysis and positional cloning of genes associated with hypertension found in populations with West African origin.

Field Center – Genetics of Hypertension  
Funder: National Heart, Lung, and Blood Institute  
PI: Richard Stanley Cooper  
Loyola University serves as one of several field sites in the Family Blood Pressure Program that has collected more than 10,000 genetic samples for analysis.

Northwestern University & Affiliates

Pancreatic Cancer Family Registry  
PI: Randall Brand  
This project serves as a repository for sociodemographic, dietary, environmental, clinical, and family history data collected from individuals and interested family members with a history of pancreatic cancer.

NUGene Project  
Funder: Center for Genetic Medicine, Northwestern University  
PI: Rex Chisholm  
This large scale gene bank is attempting to store genetic samples and associated health information from 100,000 individuals.

Molecular Genetics of Schizophrenia  
Funder: National Institute of Mental Health  
PI: Pablo V. Gejman  
This project will collect 507 schizophrenia-affected sibling pairs and will complete a genome linkage analysis.
MyGenerations
Funder: Private Donations
PI: Suzanne M. O’Neill
This project establishes three interactive computer kiosks for individuals to input family history information into; they receive scientifically determined risk assessment.

The National Ovarian Cancer Early Detection Program
Funder: The National Cancer Institute
Directors: Lee Shulman & Diljeet Singh
This program provides screening treatments that can include genetic testing and counseling for women at increased risk for ovarian cancer.

PI: Wendy S. Rubinstein
This study provides periodic MRIs for women who meet criteria for increased genetic risk of breast cancer.

Evaluation of a Family History Tool for Health Promotion and Disease Prevention
Funder: Centers for Disease Control
CO-PI: Wendy S. Rubinstein and Suzanne M. O’Neill
This study involves a controlled clinical trial of a family history screening tool developed at the CDC.

Rush University

Neurological Phenotype in FMRI Premutation Carriers
Funder: National Institute of Neurological Disorders and Stroke
Co-I: Elizabeth Berry-Kravis
Study examines neurological symptoms of carriers of faulty gene for Fragile X Syndrome.

Sickle Cell Disease Association of Illinois

Sickle Cell Disease and Newborn Screening Program Grant
Funder: Maternal and Child Health Bureau, Health Resources and Services Administration
Director: Valerie Beckley
This grant establishes a working relationship between the Sickle Cell Disease Association of Illinois and the Illinois Department of Public Health to prevent positive sickle cell newborn screens from becoming lost to follow up. A variety of community and clinician outreach activities are included.

Southern Illinois University

Biotechnology, Genetics, and Ethics Task Force
This task force, comprised of several faculty at the SIU School of Medicine, made genetics-related curriculum recommendations to the School in 2002. Currently inactive.

University of Chicago & Affiliates

IBD Genetics Consortium Site & Data Coordinating Center
Funder: National Institute of Diabetes and Digestive and Kidney Diseases  
PI: Judy H. Cho  
This grant establishes a data clearinghouse at the University of Chicago for genetic research of Inflammatory Bowel Disease.

**Multidisciplinary Psychiatric Genetics Training Program**  
Funder: National Institute of Mental Health  
PI: Eliot S. Gershon  
Receives federal funding to establish a postdoctoral training program in the genetics of psychiatry.

**Chicago Childhood Diabetes Registry**  
Funder: National Institute of Diabetes and Digestive and Kidney Diseases  
PI: Rebecca B. Lipton  
Collects cases of childhood diabetes with an eye towards testing hypotheses of familial aggregation of risk factors.

**Genetics of Breast Cancer in Blacks**  
Funder: The National Cancer Institute  
PI: Olufunmilayo I. Olopade  
Comparative BRCA 1/2 study involving 1000 West African (Nigerian) women, 360 African Americans and past research on BRCA 1/2 mutations in women of European heritage.

**Department of Human Genetics Seminar Series**  
The Department of Human Genetics at the Pritzker School of Medicine sponsors an ongoing, periodic seminar series with national scholars of genetic science and medicine.

**University of Illinois at Chicago**

**Parent's Interpretation and Use of Genetic Information**  
Funder: National Human Genome Research Institute Grant  
PI: Agatha M. Gallo  
Study examines how parents of children with a genetic condition manage genetic information and how genetic professionals help them manage this information.

**Clinical Versus Experiential Views of Genetic Disability**  
Funder: National Human Genome Research Institute Grant  
PI: Carol J. Gill  
This project compares/contrasts views on living with genetic disabilities of medical genetics professionals with views of persons living with genetic disability and the families of persons living with genetic disabilities.

**Multi-Institution Projects**

**Collaborative Genetic Study of Bipolar Disorder**  
Funder: National Institute of Mental Health  
PI: Eliot S. Gershon (University of Chicago); William A. Scheftner (Rush University)  
Collects genetic samples from 5000 individuals with bipolar disorder and 2000 genetics samples of parents with children with bipolar disorder. Multi-site study.