

State of Illinois

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# ILLINOIS STATE GENETIC SERVICES PLAN 2007

**The development of the Illinois State Genetic Services Plan involved collaboration among a variety of dedicated individuals who committed their time, energy and expertise to this effort.**

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## PLAN SUMMARY DEVELOPMENT

As more information becomes known about the importance of genetics and genomics for individual and population health, there is an increasing need to plan for the future of genetic health care and services in Illinois. In response to this need, the Illinois Department of Public Health initiated and supported a process to develop the first Illinois State Genetic Services Plan (ISGP). The plan was developed based on data collected in the Illinois Genetic Services Needs Assessment, which was conducted from 2003 through 2005; review of other state genetic services plans and other available resources; and the knowledge and expertise of many diverse participants and stakeholders. The plan development process involved the guidance of a steering committee and input from seven work groups, planning conference participants and statewide community forum attendees.

## IMPACT OF GENETIC CONDITIONS AND GENOMICS

Genetic conditions, and diseases with a genetic component, significantly contribute to morbidity and mortality in the United States. These conditions can affect human health at any stage of life; prenatally, in childhood, adolescence and in adulthood. They also have an impact on reproductive decision-making. More recently, genetics, public health and primary health care providers are becoming involved in genomics: the study of all the genes in a person, the interactions of these genes with each other and the interactions of these genes with a person's environment resulting in health or disease. It is now well recognized that, although individuals inherit genes that increase risk or susceptibility of adult chronic illnesses, such as cardiovascular disease, cancer and diabetes, the onset of these conditions can be significantly influenced by the individual's lifestyle choices. The knowledge that positive lifestyle changes will improve the overall health of individuals, especially those with a family history of such diseases, should encourage public health efforts at all levels (national, state and local) to raise public awareness in this important area. Ultimately, the overall impact of genomics integration in health care will be to improve health by reducing morbidity and mortality through coordinated disease prevention and health promotion efforts.

## PRIORITY ISSUES: GOALS AND OBJECTIVES

The Illinois State Genetic Services Plan addresses seven key priority areas that were identified through the state needs assessment process and broad stakeholder input. The following goals, objectives and action items provide the foundation for the state genetic services plan. The order of the objectives and action items for each goal are prioritized.

### GOAL 1: Promote high quality, comprehensive and accessible genetic services for all Illinois residents.

Genetic services are available to help individuals and families understand their specific genetic condition, or risk of genetic condition, and ultimately improve health by reducing morbidity and mortality associated with these conditions. Due to advances in genetic technology and increasing clinical applications for more common conditions, the need for genetic services is growing. Comprehensive genetic services include evaluations, testing, counseling, treatment, management, educational activities, family support and follow-up for all members of the population. Assuring quality and accessible genetic services to the public is a fundamental component of the state genetic services plan.



### OBJECTIVE 1

**Evaluate and improve the availability and accessibility of genetic services in Illinois.**

#### ACTION ITEMS

1. Regularly review select components (*includes locations, referral patterns, numbers of patients seen*) of the Illinois Department of Public Health genetics grantees.
2. Evaluate genetic services delivery in Illinois (*Action Item 1 components and other*) and identify gaps (*collaborate with Data and Public Health Infrastructure efforts, Objective 1, Action Item 1*).

3. Monitor the Department's Newborn Screening Program in order to identify, evaluate and incorporate, when appropriate, new technologies to facilitate screening and follow-up.
4. Expand the Department's laboratory and follow-up program capacity to support comprehensive and emerging testing, tracking, and treatment options for genetic conditions.

## OBJECTIVE 2

**Create and support a Genetic Services Network to share strategies for providing quality, accessible and comprehensive services to all residents.**

### ACTION ITEMS

1. Recruit network members, including representatives from the Department's genetic grantee sites.
2. Develop mission, goals, structure, desired outcomes and administration of the Genetic Services Network.
3. Identify existing protocols or develop guidelines describing appropriate genetic/genomic services.
4. Develop strategies for providing accessible and comprehensive services (*see Barriers to Access to Care Objectives 1, 2, 3 and 4*).
5. Adapt or develop quality indicators for genetic services in Illinois.
6. Publicize the network and the availability of genetic services and related support services to all residents .
7. Evaluate the network outcomes.



## **GOAL 2: Reduce barriers to access to genetic health care services.**

Advances in genetic technology and genomics have led to the development of primary, secondary and tertiary prevention strategies. However, access to genetic services is not equally available and utilized by all segments of the population, particularly in underserved rural, low-income and minority communities. Understanding and reducing potential barriers to access to genetic services is key in providing equal opportunities for disease prevention and health promotion.

### **OBJECTIVE 1**

**Improve access for medically underserved individuals by addressing economic barriers.**

#### **ACTION ITEMS**

1. Raise public and professional awareness of available sources of reimbursement for and coverage of genetic services (collaborate with Financing and Reimbursement efforts, Objectives 1 and 2).
2. Link clients to available genetics advocacy and consumer support groups.

### **OBJECTIVE 2**

**Improve access for medically underserved individuals by addressing geographic barriers.**

#### **ACTION ITEMS**

1. Evaluate genetic services delivery and identify gaps (collaborate with Genetic Services efforts, Objective 1, Action Item 2).
2. Examine the need for establishing additional genetics outreach clinics in underserved areas.
3. Identify and increase funding opportunities for providing genetic services in underserved areas.
4. Review existing genetics telemedicine efforts and conduct a study to assess the feasibility of telemedicine for genetic services in rural areas.
5. Link clients to health departments for assistance with transportation to nearest genetics clinics.

## OBJECTIVE 3

**Improve access for medically underserved individuals by addressing language and cultural barriers.**

### ACTION ITEMS

1. Assess cultural barriers to receiving genetic services.
2. Identify groups who would benefit from interpreter services.
3. Determine methods of providing interpreter services to those in need, including raising awareness of existing services and costs of those services.
4. Evaluate current genetics education literature for cultural competence and develop resources where there is a gap in existing materials (collaborate with Public Education efforts, Objective 2, Action Item 2).



## OBJECTIVE 4

**Improve access for medically underserved individuals by addressing educational barriers.**

### ACTION ITEMS

1. Collaborate with Public Education efforts, Objectives 1, 2 and 3.
2. Collaborate with Professional Education efforts, Objectives 1 and 2.

## **GOAL 3: Improve public health data and infrastructure to support and advance genomics activities.**

Developing a state genomics infrastructure requires invested leadership, genomics expertise and accessible genetics data. This infrastructure is critical to the successful integration of genomics and related technologies into health promotion and prevention activities, disease management, and other areas throughout the public and private health care systems.

### **OBJECTIVE 1**

#### **Develop a public health infrastructure to support genomics activities.**

##### **ACTION ITEMS**

1. Examine the effectiveness of grants to genetic service providers and local health departments for genetic screening and outreach (collaborate with Genetic Services efforts, Objective 1, Action Item 2).
2. Continue efforts to integrate genomics into other program areas (e.g., chronic disease, cancer, infectious disease) and across the lifespan.
3. Educate and raise awareness of public health policymakers and agency leaders about the importance of genomics (collaborate with Public Education efforts, Objective 3).
4. Justify and identify funding for additional staff for data/epidemiology, education and incorporating genomics into chronic disease programming.
5. Identify and develop partnerships and resources that may support the implementation of the Illinois State Genetic Services Plan.



## OBJECTIVE 2

**Improve the accuracy and availability of data regarding genetic conditions and services.**

### ACTION ITEMS

1. Inventory existing databases and archived data, and assess ability for linkage or sharing.
2. Increase awareness of public health program staff and partners regarding the types and quality of data currently collected by and available from state agencies.
3. Improve the accuracy and completeness of the data currently collected by state agencies.
4. Make available aggregate genomics data to the public via Internet capable systems, as well as to individuals without Internet access (hard copies).
5. Make individual, de-identified public use data sets available to public health researchers and other interested individuals via internet capable systems.
6. Incorporate the Department's Genetic Screening Tool into the Cornerstone data system.



## **GOAL 4: Increase genomic awareness and literacy in the general public.**

A genomics-literate population is more likely to apply genomic information to health and lifestyle decisions and related ethical, legal and social issues. To increase genomics literacy in the population, education efforts should begin early in student education. Efforts targeted at specific community groups also can be implemented. Educators, policymakers, health care providers, spiritual leaders and the media are all key partners in public genomics education efforts.

### **OBJECTIVE 1**

**Improve genomics knowledge and education in the primary and secondary school systems.**

#### **ACTION ITEMS**

1. Identify and educate stakeholders (e.g., legislators and administrators) in the state education system.
2. Partner with the Illinois State Board of Education to review and enhance existing genetics/genomics components of the Illinois Learning Standards.
3. Identify existing genomics curriculum materials, adapt or develop necessary materials and make them easily available to educators.
4. Provide genomics education for educators, both existing and currently in training programs.



## **Increase genetic awareness and knowledge in all segments of the population.**

### **ACTION ITEMS**

1. Develop a comprehensive, culturally sensitive Web site to provide information and resources about genomics to the public.
2. Identify leaders in underserved and minority communities for the purpose of building relationships and disseminating information about genomics and public health.
3. Identify and develop appropriate written materials for individuals without Internet access.
4. Develop and coordinate a statewide Genetics Awareness Day, in coordination with existing national DNA and family history days.
5. Identify existing genetic and family history awareness programs/materials and replicate successful models and projects.
6. Identify religious professionals/spiritual leaders for the purpose of building relationships and disseminating information about genomics and public health.
7. Identify “mass media” mechanism for promoting family history.
8. Develop relationships with media contacts.



## **OBJECTIVE 3**

### **Increase genetic awareness and knowledge among state policymakers, including legislators, other elected officials and public health leadership.**

#### **ACTION ITEMS**

1. Form a coalition of partner organizations.
2. Develop a strategy for approaching state policymakers.

## **GOAL 5: Promote integration of genomics into health care delivery systems through education of health professionals and the health care work force.**

As new strategies and services evolve from genomic technologies, it will become increasingly important to provide education and training to health care professionals and the health care workforce to enhance awareness, build competencies and ensure integration of genomics in the health care delivery system. Health care providers' current level of awareness and understanding of genetic conditions and genomics has not kept pace with the current increase in genomic knowledge. Therefore, by increasing provider awareness and knowledge of genomics, health care professionals may make more informed decisions regarding delivery of genetic services.

### **OBJECTIVE 1**

**Identify the educational needs and assess the current state of genomics knowledge of the public health and health care work force.**

#### **ACTION ITEMS**

1. Identify existing genomics competencies for health professionals.
2. Identify liaisons and develop relationships with state professional organizations.
3. Review published studies assessing health professional needs and knowledge in genomics to provide direction for educational efforts.
4. Pending results of Action Item 3, conduct a formal research study to assess genomic knowledge and integration of genomics into medical practice among health professionals and the health care workforce.

### **OBJECTIVE 2**

**Increase health professionals' genomics knowledge through identification, development and dissemination of educational resources.**

#### **ACTION ITEMS**

1. Identify existing genomics educational resources for health care providers.
2. Assess and evaluate the quality of available genomics resources for health care providers and identify gaps in educational materials.
3. Create a clearinghouse of genetics educational resources for health care professionals, accessible via the Department's genetics program Web site, which would include curricular materials, referral resources and guidance, a speaker's bureau with list of speaker topics, and other pertinent information.

4. Disseminate and raise awareness about clearinghouse materials (Action Item 3) to the health care workforce through relationships developed under Objective 1.
5. Develop regular and ongoing state wide CME/CEU opportunities for continuing genomics education and updates.
6. Develop genomics curricular materials where there is a gap in available educational resources.

## OBJECTIVE 3

### Evaluate the effectiveness of Objectives 1 and 2.

#### ACTION ITEMS

1. Assess referral patterns of primary care providers to genetic service providers, including the appropriateness of referrals and “missed opportunities” for referrals.
2. Monitor usage (“hits”) of the Department’s clearinghouse Web site.
3. Survey health care professionals regarding changes in their genomics knowledge (baseline is Objective 1, Action Items 3 and 4).
4. Identify other appropriate outcomes and measures to evaluate Objectives 1 and 2.

## OBJECTIVE 4

### Assess and assure an adequate work force supply and access to genetic providers.

#### ACTION ITEMS

1. Conduct ongoing surveys, or obtain existing data from the Illinois Genetic Services Needs Assessment, to determine supply and distribution of qualified genetic providers.
2. Develop and support undergraduate pipeline programs and graduate medical education programs that will lead to an adequate and diverse genetic provider workforce.
3. Assure board certification and recertification as appropriate for genetic providers.
4. Partner with genetic professionals and health systems to investigate opportunities for providing genetic consultation and counseling through electronic communications networks, thus extending genetic clinical services to areas currently without such access.
5. Convene a study group to investigate alternative genetic service delivery models, including use of other types of health care providers.

## **GOAL 6: Address genetic services financing and reimbursement issues that impact individuals, families, and/or genetic service providers.**

Adequate financing and reimbursement for genetic services by public and private sources can significantly impact the genetic services delivery system. Limited insurance coverage and reimbursement may reduce access to and availability of genetic evaluations, counseling, tests and treatments. As many public and private financing decisions are made at the state level, it is important to have the necessary data to improve coverage and reimbursement for genetic services in public genomics education efforts.

### **OBJECTIVE 1**

**Assess current status of reimbursement for clinical genetic services.**

#### **ACTION ITEMS**

1. Identify existing efforts to study and gather data on reimbursement for genetic services, nationally or in other states.
2. Survey genetic service providers regarding reimbursement for genetic services to gather data on the following:
  - a. Clinical genetic services activities that are part of evaluation, testing, management and counseling.
  - b. CPT and other codes used for the above activities.
  - c. Proportion of patients by payer type (such as Medicaid, Medicare, private insurance) and service category (including preconception, prenatal, pediatric, adolescent, adult).
  - d. Reimbursement for different activities/CPT codes by payer type.
  - e. Reasons for non-payment by different payers.
  - f. Identify gaps in services provided, billed for and reimbursement.
3. Examine existing public and private insurance policies regarding coverage and reimbursement for genetic testing, screening, and services.
4. Obtain reimbursement rates and policies regarding genetic services from the state's Medicaid program.
5. Conduct cost-effectiveness studies regarding coverage for genetic health services including estimates of changes in state and private expenses if additional services are covered.

## OBJECTIVE 2

**Based on the results of Objective 1, explore strategies for improving third-party coverage and reimbursement for genetic health services.**

### ACTION ITEMS

1. Identify liaisons with third-party payers and the state's Medicaid program.
2. Based on data gathered in Objective 1, advocate for improved private insurance coverage of genetic services by identifying or developing a presentation or other materials to educate private health insurers about the value, financing, delivery and organization of genetic services.
3. Based on data gathered in Objective 1, advocate for improved Medicaid coverage of genetic services by identifying existing models and developing materials to educate Medicaid policy makers about the value, financing, delivery and organization of genetic services.
4. Educate genetic and other specialty providers about optimal billing and coding practices, using existing educational resources when available, and developing new materials as needed.

## OBJECTIVE 3

**Examine billing by genetic counselors for the provision of genetic services.**

### ACTION ITEMS

1. Conduct study to assess how genetic counselors are currently billing.
2. Examine genetic counselor billing practices in other states.
3. Identify the necessary steps, as well as the advantages and disadvantages, regarding implementation of genetic counselor billing.
4. Coordinate state efforts with national organizations.



Photo courtesy of the University of Maryland Medical Center ([www.umm.edu](http://www.umm.edu))

## **GOAL 7: Identify and examine the ethical, legal and social issues (ELSI) relevant to clinical genetic services, genetic research and related applications.**

Ethical, Legal and Social Issues (ELSI) are critical to all facets of the Illinois State Genetic Services Plan. The ELSI goal aims to address existing and anticipated issues that likely will develop as a result of technological advancements in genomics. Consideration of ELSI in the plan is necessary to establish ethical and legal standards that will protect and promote the interests of individuals, families and communities throughout the state.

### **OBJECTIVE 1**

**Establish an ELSI advisory group, consisting of diverse professional and public members, to advise the Illinois Department of Public Health regarding ethical, legal and social issues relevant to clinical genetics and genetic research.**

#### **ACTION ITEMS**

1. Review and document other states' legislation and policies related to informed consent for newborn screening.
2. Identify informed consent issues related to prenatal, preimplantation and preconception genetic diagnosis, genetic testing of children, genetic testing for competent adults and population-based genetic screening.
3. Identify informed consent and other issues related to genetic research.
4. Review and develop access and usage guidelines related to stored tissue samples, including biobanks.
5. Review and document genetic privacy legislation and regulations, and identify any cases of genetic discrimination in employment or insurance.
6. Develop a Genetic Bill of Rights.
7. Serve as a resource to review pending state legislation related to clinical genetics and genetic research.
8. Address social justice issues related to advances in genetics, particularly issues related to health disparities, religious concerns, cultural differences and vulnerable populations.



## FUTURE EFFORTS

The Illinois State Genetics Services Plan is intended to serve as a framework for guiding future directions in genetic service-related activities. The state plan efforts can be used to mobilize individuals, organizations, institutions, communities and policy makers interested in improving access to, and outcomes of, genetic health care. Effective implementation, however, will require ongoing, coordinated collaboration of invested stakeholders and partners. The following are anticipated activities for implementation of the Illinois State Genetics Services Plan:

- Disseminate the plan widely throughout the state.
- Begin plans to implement specific action items within three months of state plan publication.
- Identify individuals and/or organizations to conduct activities that will carry out the mission of the state plan goals, objectives and action items.
- Identify, coordinate and secure funding opportunities for implementation efforts.
- Facilitate new and existing partnerships and collaborations.
- Develop and administer an annual evaluation to review goal attainment.
- Report evaluation and progress to stakeholders and seek continued input.



