Goal 2 Public Health Genetics Infrastructure

Develop and maintain a responsive public health genetics program to plan, implement, monitor, and evaluate genetics education and services in Oklahoma.

Through the efforts of the Human Genome Project, genetics, once an obscure program for rare genetic disorders, will soon become an integral part of our health care system and provide amazing opportunities for disease prevention and health promotion. Today, public health genetics is defined as the application of advances in genetics and molecular biotechnology to improve public health and prevent disease (Khoury, Genetics n. pag.).[28] Historically, public health's established network of essential services has successfully worked to prevent diseases, provide early screening, promote and protect health, and assure access to the health care system. This network can now facilitate the integration of genetic advances into the health care system and into public health practice. Through public health core functions of assessment, policy development, and assurance, state public health agencies must, among other activities, ensure genetic services are integrated into the health care system and are accessible and utilized to benefit the health outcomes of citizens. The *State Genetics Plan* will provide the guidance to ensure Oklahoma benefits from genetic advances through the establishment of a collaborative, comprehensive public health genetics program to assure an infrastructure exists to plan, implement, monitor, and evaluate genetics in Oklahoma. Public health must proactively develop an infrastructure capacity to ensure: (1) genetic advances are incorporated into public health practice; (2) local clinical genetic services are maintained, accessible, meet standards, and have the capacity to meet the needs of all Oklahomans; (3) citizens are knowledgeable and can advocate for genetic services to improve their health; (4) providers have the tools needed to integrate genetics into their daily practice; and (5) systems exist to monitor the ethical, legal, and social issues related to local genetic services. The *State Genetics Plan* for infrastructure was developed by reviewing information and documents from the Human Genome Project, Healthy People 2010 initiative, All Aboard the 2010 Express: *A Ten-Year Action Plan for Children with Special Health Care Needs and Their Families,* publications from the Council of Regional Networks for Genetic Services, public health data system plans to create a Child Health Information System, and the statewide genetic needs assessment findings related to reimbursement for genetic services. The following segment for goal 2 will provide a brief review of these resources and an overview of the evaluation activities for the plan. The action steps for Public Health Genetics Infrastructure, Goal 2, begin on page 43.

A public health genetics program in the future will be as important in disease prevention as immunization **programs are today.**

[28] *Genetics and Public Health...*

Human Genome Project

The Human Genome Project is the science that is thrusting public health into the molecular age of health maintenance. The Human Genome Project began in 1990 as a collaborative project between the U.S. Department of Energy and the National Institutes of Health (NIH) with the goal to map and sequence the human genome. Today, several types of genome maps have been completed, and a working draft of the entire human genome sequence was announced in June 2000 (Human Genome 1).[25] Oklahoma has played a role in this exciting project through Dr. Bruce Roe's efforts to successfully sequence chromosome number 22. For more information about local efforts, visit this Web site: www.genome.ou.edu. The ultimate goal of the Human Genome Project is to "use this information to develop new ways to treat, cure, or even prevent the thousands of diseases that afflict humankind" (Medicine 1).[38] Since virtually all disease, except some cases of trauma, have a genetic component (Collins and McKusick 540),[8] the opportunities to improve the health of Americans are staggering. The anticipated paradigm shift from illness care to preventive medicine interfaces with public health's long history in health promotion and disease prevention. It is predicted that a public health genetics program in the future will be as important in disease prevention as immunization programs are today.

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Healthy People 2010 and Children with Special Health Care Needs - 2010 Express

The overarching goals of the Healthy People 2010 initiative of the US Department of Health and Human Services are to "increase quality and years of healthy life" and "eliminate health disparities" (Healthy 1).[24] Technological advances in the field of genetics hold great promise to meet this challenge and fulfill the 2010 goals and purpose of promoting health and preventing illness, disability, and premature death. To facilitate the attainment of Healthy People 2010, the public health genetics program developed a comprehensive plan to address genetics from infancy to adulthood.

Although genetics impacts the entire lifecycle, the children with special health care needs (CSHCN) population will be a target population for special monitoring by the OSDH genetics program. The *All Aboard the 2010 Express* document defines CSHCN "as those children who have or are at increased risk for a chronic physical, developmental, behavioral, or emotional condition and who also require health and related services of a type or amount beyond that required by children generally." National estimates of the number of children with special health care needs range between 15 and 20 percent of all children (U.S. Department 4).[53] Of the CSHCN population, at least 10% have chromosomal problems

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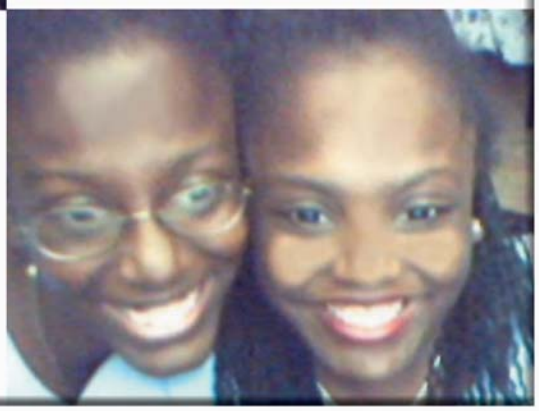
[25] *Human Genome Project…*
[38] *Medicine and the New….*
[8, 8.2] *Implications...*
[24] *Healthy People...*
[53] *US Department of Health…*

and 3% have genetic related disorders. However, considering the genetics of diabetes, asthma, cancer, and hearing loss, clearly a significant percentage of children with special health care needs have conditions with a genetic component (Cunningham, Percent n. pag.).[11] To ensure the CSHCN with genetic disorders receive needed services, the state plan outlines the development of strong links to all programs that serve CSHCN. To promote and facilitate the development of community-based, family-centered, culturally competent, and coordinated systems of care, the *2010 Express* document's six core outcomes were utilized to develop the plan's service systems for CSHCN.

The genetics program will build on the successful newborn screening program follow-up systems to enhance service systems for infants identified through public health screening programs into adulthood, with the ultimate goal to expand these service systems to all CSHCN affected by genetic disorders. The establishment of medical homes with access to needed services will be a key performance measure of the public health genetics program. A medical home is a source of routine healthcare in the community that assists in early identification, provides ongoing primary care, and coordinates with a broad range of other special, ancillary and related services. The state plan outlines a community-based approach to develop an infrastructure that operates across service sectors promoting program communication and collaboration to better meet the needs of the child and the family. The planned activities to achieve success with the *2010 Express* six core outcomes are outlined throughout the *State Genetics Plan's* action plan. Children with special health care needs in Oklahoma will benefit from the successful implementation of the plan.

The Smith sisters have a common genetic disorder prevalent in African Americans.

Kayla and Dewayna's story can be found on page 54.



[11] *Cunningham, George...*

A summary of the six core outcomes of *All Aboard the 2010 Express* and how the OSDH Genetics Program and implementation of the *State Genetics Plan* will facilitate achievement of these core outcomes:

All Aboard the 2010 Express is a 10-year action plan with six core outcome measures. The following identifies each core outcome and the related genetic program and *State Genetics Plan* activities (U.S. Department 7):[54]

1. Families of children with special health care needs will partner in decision making at all levels and will be satisfied with the services they receive.

Genetics Program: Families are currently represented on the Oklahoma Genetics Advisory Council and on each OGAC committee. A Family Advisory Committee of OGAC has been established.

2. All children with special health care needs will receive coordinated, ongoing, comprehensive care within a medical home.

State Plan: A long-term case management follow-up program will initially be established for the Newborn (metabolic and hearing) Screening Program and later expanded to include all CSHCN affected by genetic disorders.

3. All families of children with special health care needs will have adequate private and/or public insurance to pay for the services they need.

State Plan: Public health leadership and advocacy activities in collaboration with OGAC and its committees and planned case management services will assist with this outcome.

4. All children will be screened early and continuously for special health care needs.

State Plan: Public health genetic and newborn screening infrastructure development, including the maintenance of short-term follow-up activities, and planned long-term follow-up case management system and data integration projects, will provide screening and effective referral for services.

5. Community-based service systems will be organized so families can use them easily.

State Plan: Public health genetic and newborn screening infrastructure development and collaboration with OGAC and its committees will develop effective systems to serve CSHCN.

6. All youth with special health care needs will receive the services necessary to make transition to all aspects of adult life, including adult health care, work and independence.

State Plan: The planned collaborative project with the OUHSC to establish an Adult Transition Program by establishing a system to serve sickle cell disease and cystic fibrosis clients and later expanded to include all CSHCN affected by genetic disorders will facilitate attainment of this outcome.

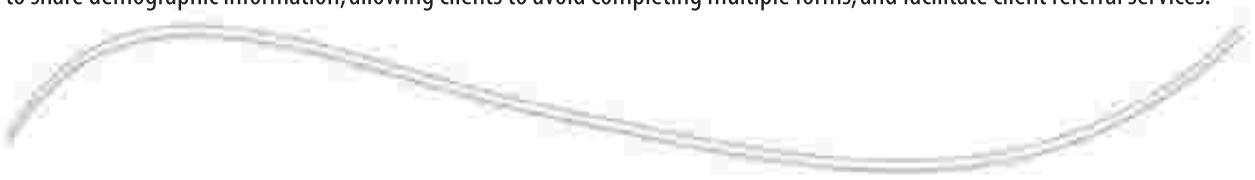
[54] *US Department of Health…*

Council of Regional Networks for Genetic Services (CORN)

In 1985, the Council of Regional Networks for Genetic Services (CORN) was formed through funding by Maternal Child Health Bureau, Health Resources and Services Administration. CORN was established to provide a forum for dialogue and national coordination among the U.S. genetic networks. In 1997, the CORN published guidelines for the development of a public health genetics program. The purpose of the guidelines was to provide state and public health agencies with an outline of suggested components for a genetic services system. CORN identified standards for genetic services and charged public health to participate in quality assurance measures to ensure all populations benefit from genetic services. Through assessment, policy development, assurance, and collaboration, state genetic programs can prevent morbidity and mortality throughout the lifecycle (CORN 14).[10] The CORN guidelines provided a useful framework in developing the action plan. CORN no longer exists today, but collaboration between states continues through the National Newborn Screening and Genetics Resource Center (NNSGRC). The NNSGRC Web site is at genes-r-us.uthscsa.edu.

Integration of Public Health Data Systems

The integration of public health data systems is an important component to the development of an infrastructure with the capacity to monitor and evaluate genetic services. The *State Genetics Plan* addresses data integration, and many projects are underway to develop an OSDH Child Health Information System (CHIS). The CHIS project has two purposes: (1) allow health care providers to access a child health profile to ascertain an infant's screening and immunization status, and (2) allow effective referral and monitoring; is the child enrolled in Early Intervention, WIC, etc? Current integration activities include the integration of three public health data systems: (1) newborn hearing, (2) newborn metabolic disorder screening program, and (3) vital records. A Newborn Hearing Screening federal grant is funding this integration project. After this project is successfully implemented, the genetics program will participate in assessing the feasibility of integrating with immunization and other public health program databases, such as WIC and Lead Screening. Additional integration efforts will involve the OSDH county health department data system PHOCIS (Public Health Oklahoma Client Information System). PHOCIS provides an overview of the services provided to each citizen by the health department. Linking with this system will provide oversight capabilities; did a child obtain Early Intervention services? Broader integration efforts will be explored and include possible linkage with the Joint Oklahoma Information System Network (JOIN) effort. JOIN is a statewide initiative to link agency information systems to avoid the duplication of data by allowing agencies to share demographic information, allowing clients to avoid completing multiple forms, and facilitate client referral services.



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[10] *Council of Regional . . .*

Reimbursement for Genetic Counseling, Testing and Newborn Screening

The issue of reimbursement for genetic counseling and genetic tests, including the genetic screening provided by the OSDH newborn screening program, was identified as a potential barrier to genetic services during the genetics needs assessment conducted by OSDH in June 2001. This issue has been recognized as a problem nationally. Genetic counseling is an essential service to ensure individuals and families understand the implications and limitations of genetic testing. In most cases, genetic testing does not predict absolutely whether an individual will manifest a condition later in life, with the rare exception of single gene disorders such as Huntington disease. Rather, a positive result indicates the increased likelihood of illness based on an individual's genetic makeup. Board-certified genetic counselors are individuals trained to effectively communicate information about the heritability of disorder/disease, available genetic testing, screening, prognosis, and recurrence within a framework of understanding and support. However, access to a board-certified counselor is limited to the physicians who are committed to providing genetic counseling despite the lack of reimbursement for such services. In the United States, there are no state or federal laws requiring insurers to offer or cover genetic counseling (Johnson 2).[27]

For genetic testing, local genetic laboratories reported insurance providers often do not acknowledge them as a reimbursable provider. Therefore, the geneticist is often forced to utilize an out-of-state lab, unless the patient is willing to pay for the genetic test. This can result in delayed receipt of test results, and can inhibit consultation between ordering geneticists and the laboratory clinical experts, which is necessary to facilitate the diagnostic process. For the public health newborn screening program, the fee has been inadequate to cover current testing and is the limiting step in the expansion of screening, such as Medium Chain Acyl-CoA Dehydrogenase Deficiency (MCAD), that could reduce morbidity and mortality in children. The plan addresses issues of reimbursement through collaboration with OGAC and its committees, insurance providers, stakeholders, and the genetics program.

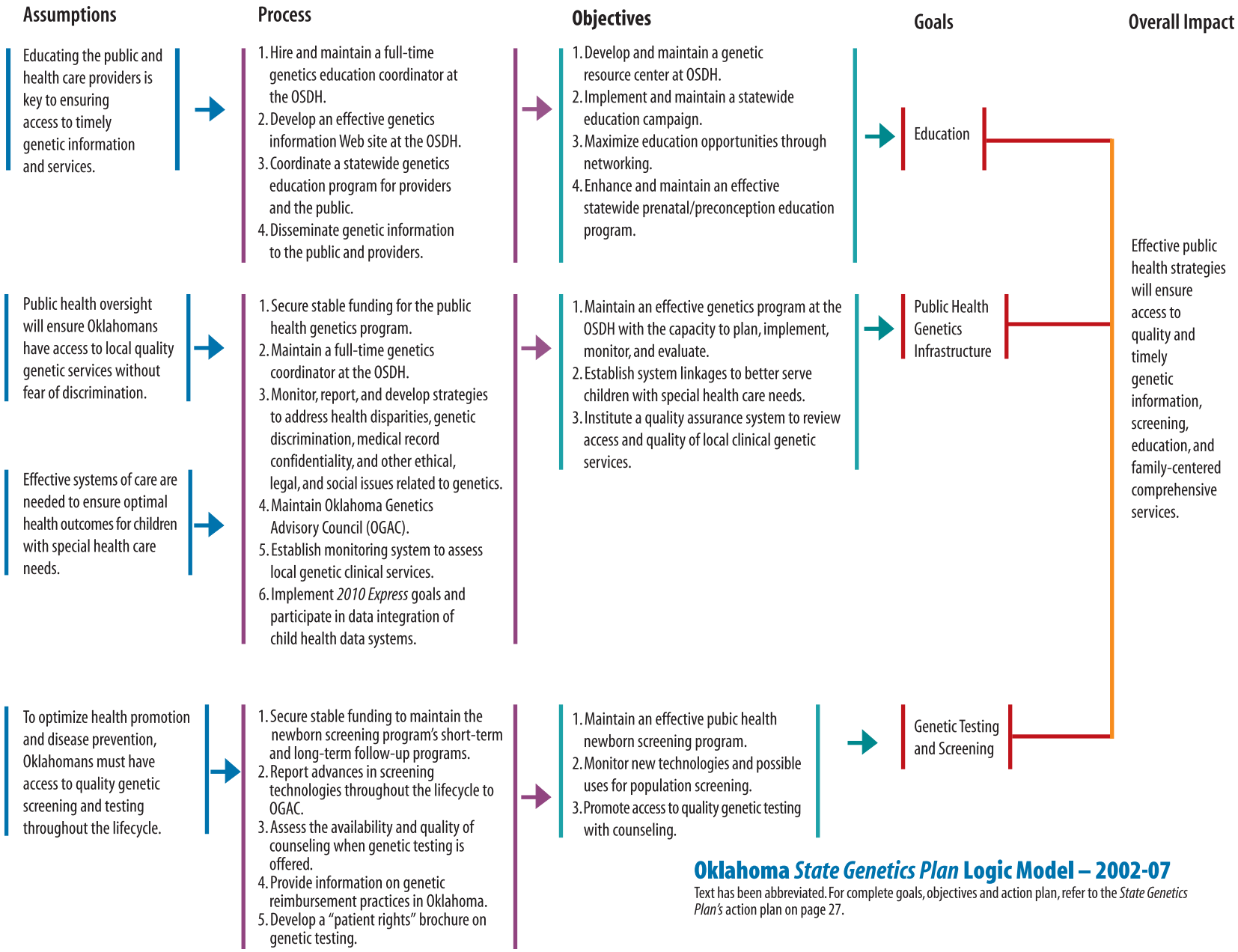
Evaluation

Evaluation will be an essential component of the public health genetics program. To determine if the OSDH genetics program is achieving the mission of the state plan, a comprehensive assessment and evaluation system will be established. The Institute of Medicine recommends every public health agency regularly and systematically collect, assemble, analyze and make available information on the health of the community, including statistics on health status, community health needs and epidemiologic and other studies of health programs (Khoury and Genetics 1719).[29] A logic model (page 42) of the state genetic plan has been developed. Utilizing the logic model and with the collaboration of the Evaluation Committee of OGAC, a comprehensive evaluation plan will be established to monitor goals and mission. Performance measures and outcomes will be clearly defined and monitored. Mechanisms will be established to track whether or not planned project activities (process) were actually carried out. Standardized data tools will be developed and incorporated into current data assessment programs such as Pregnancy Risk Assessment Monitoring System (PRAMS), and The Oklahoma Toddler Survey (TOTS). PRAMS and TOTS are OSDH population-based surveillance systems that provide health data on women and toddlers in Oklahoma. Data tools will be developed for the six core outcomes of 2010 Express and for related Title V performance measures. Effectiveness of the attainment of goals and mission will be monitored and assessed by the genetics program in collaboration with OGAC. The *State Genetics Plan's* action steps will be tailored as needed to efficiently and effectively achieve the mission.

[27] *Johnson, Alissa. . .*

[29] *From Genes to Public Health. . .*

Genetics Program of the Oklahoma State Department of Health



Oklahoma *State Genetics Plan* Logic Model – 2002-07

Text has been abbreviated. For complete goals, objectives and action plan, refer to the *State Genetics Plan's* action plan on page 27.

Summary

Goal 2

Through participation of the Oklahoma Genetics Advisory Council (OGAC) and its committees a comprehensive representative action plan for goal 2 was developed. The *State Genetics Plan's* three objectives for goal 2 include: (1) public health genetic infrastructure development, (2) system linkages to better serve children with special health care needs (CSHCN) population, and (3) clinical genetic services. The combined Public Health Policy and Evaluation Committee of OGAC identified five action steps as priority for implementation.

Goal 2 Action Plan

"Develop and maintain a responsive public health genetics program to plan, implement, monitor, and evaluate genetics education and services in Oklahoma."

Objective 1- The OSDH will develop and sustain a responsive, collaborative, culturally sensitive, and effective public health genetics program that links with community partners and addresses health disparities.

Action Steps (◇ symbol identifies prioritized action steps):

- ◇ Identify stable funding sources for the public health genetics program (priority number 2).
- ◇ In collaboration with Oklahoma Genetics Advisory Council (OGAC), establish a network to address and monitor local ethical, legal, and social issues related to genetics including confidentiality and discrimination, e.g., life insurance discrimination based on genetic testing, inflated insurance premiums due to a diagnosis of a genetic disorder, or Medicaid eligibility (priority number 3).
- ○ Establish a system for the public to report incidents of insurance and employee discrimination related to genetics.
- ○ Maintain a full-time State Genetics Coordinator at the OSDH to provide oversight of the implementation of the *State Genetics Plan* and to administer the public health genetics program.