Goal 3 Genetic Testing and Screening

Maximize the quality of genetic testing and the effectiveness of public health screening programs to serve all the citizens of Oklahoma.

One of the first clinical applications emanating from the Human Genome Project is the rapidly expanding use of genetic tests. "Already more than 500 genetic tests have been developed and hundreds are in the pipeline that may diagnose disease or identify disease risk before symptoms occur" (Slater 2).[51] Genetic testing presents complex social issues. Unlike other traditional laboratory tests, genetic testing provides opportunities to identify persons with genetic diseases, and/or carriers of genetic disease (not affected, but could pass on to future generations), and identify individuals at risk for possible, but not definite disease conditions in the future. Genetic tests not only provide intimate information about the patient being tested, but also the family. With the sequencing of the human genome, molecular (DNA) genetic testing will only continue to evolve to assist in the identification of patients and families at risk for specific diseases. However, safeguards must be in place to ensure genetic testing is performed in an environment that informs the client of the limitations, implications, and benefit. In addition, safeguards must exist to prevent employment and insurance discrimination, and breaches in confidentiality. The following segment for goal 3 will provide a brief review of the Secretary's Advisory Committee on Genetic Testing recommendations, and a review of the success and challenges of the population-based genetic screening program model, the Oklahoma Newborn Screening Program. The newborn screening public health program will be the core infrastructure to begin the development of a system with the capacity to provide public health oversight for genetic testing throughout the lifecycle. Action steps for Goal 3 begin on page 57.

Secretary's Advisory Committee on Genetic Testing

The Secretary's Advisory Committee on Genetic Testing (SACGT) was chartered in 1998 to advise the Department of Health and Human Services on the medical, scientific, ethical, legal, and social issues related to the development and use of genetic tests. Recommendations of this advisory body were released for public comment in April 2000, and formally released to the public in November 2000. The four overarching principles of the SACGT include: (1) ensure genetic testing is utilized to improve the health and well-being of individuals and families, (2) ensure individuals and families who undergo genetic testing have access to genetic education and counseling to ensure their ability to make an informed decision about being tested, (3) efforts to ensure the education of the public as well as health providers about genetics is necessary, and (4) issues of discrimination in employment and health insurance must be addressed by policymakers to prohibit such discrimination or the public will be reluctant to undergo genetic tests that might be beneficial to its health and well-being (*Enhancing* vii).[16] At the SACGT education conference, Dr. Slater eloquently summarized the issues related to genetic testing:

[51] *Genetic Testing…*

[16] *Enhancing the Oversight…*

> As the use of genetic tests expands and a broader range of health providers begin using these powerful diagnostic tools, health professionals will be faced with answering questions and requests for genetic tests from their patients. Often these patients may know very little and what they think they know may be inaccurate or, at best, incomplete. Health professionals will need to be equipped with the knowledge to understand when it is appropriate to order a test for certain patients and how to interpret and apply the test results when they are returned. They must also be able to recognize when it is appropriate to refer patients to a geneticist or other specialist. Some of the challenges posed by genetic testing for health professionals have already been documented. Several years ago, a study published in the New England Journal of Medicine reported some worrisome findings. The study assessed indications for the use and interpretation of the genetic test for the APC gene, which is responsible for familial adenomatous polyposis. In almost 32 percent of the 177 cases studied, physicians misinterpreted the test results. Only 19 percent of the patients received genetic counseling before the test, and only 17 percent provided written informed consent. In addition, many of the physicians interviewed did not recognize the limitations of the testing. As a result, patients may have undergone unnecessary testing and experienced additional stress. Some may still be undergoing unwarranted additional surveillance today (Slater 3).[52]

As addressed in goal 1, education will be an essential task for the public health genetics program. Action steps for goal 3 address the establishment of a capacity to provide public health oversight to ensure genetic testing is offered in an ethical and informed environment. Utilization of OGAC and its committees and the development of the newborn screening program's infrastructure will be the initial steps toward developing the capacity to monitor genetics throughout the lifecycle.

Newborn Screening Program

Screening at-risk populations for genetic disorders is not a new public health function. Since the sixties, state public health programs have administered newborn screening programs. The first U.S. population-based screening program for children began with testing all infants at birth for the genetic disorder phenylketonuria (PKU). This disorder is not easily diagnosed by physicians and without early identification and treatment within the first month of life, profound mental retardation occurs. Screening infants at birth for PKU prevents mental retardation through early identification and subsequent treatment. Every state has a newborn screening program. Each state selects the disorders to include in the newborn screening program. States vary in the number of disorders screened with a range as low as three to over 30; however, every state screens for the genetic disorder of PKU and for congenital hypothyroidism. In Oklahoma, all newborns have the simple heel-stick blood test to screen for phenylketonuria (PKU), congenital hypothyroidism, galactosemia, and sickle cell disease, and a hearing screen to detect infants at risk for and with hearing loss. By Oklahoma law and established rules and regulations, every newborn is required to be screened within the first week of life to ensure early identification, referral, and treatment.

The two main issues facing every state newborn screening program include the provision of comprehensive follow-up services and the expansion for additional disorders. Controversy over disparities between state-operated screening programs, and the importance of short-term and long-term follow-up programs in the prevention of morbidity and mortality associated with the disorders, has led to the development of guidelines to facilitate the development of comprehensive state public health newborn screening programs. The "U.S. Newborn Screening System Guidelines II: Follow-up of Children, Diagnosis, Management, and Evaluation Statement of the Council of Regional Networks for Genetic Services" established national newborn screening system guidelines to facilitate states in developing effective newborn screening systems. This report underscores the need for case management and service systems and specifies that newborn screening programs need a coordinated system of services for follow-up, diagnosis, and treatment of children identified with a disorder. Children with chronic and disabling conditions and their families often require an extensive range of different services. Assurances that infants identified through newborn screening with a disorder are established in a medical home is an essential component of a comprehensive newborn screening program. The ideal medical home includes a primary care physician working in partnership with families and other specialists, subspecialists, and the wide range of other providers needed to promote optimal health (Pass et. al. 1-2).[43]

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[52] *Genetic Testing…*

[43] *US Newborn Screening…*

In August 2000, The American Academy of Pediatrics (AAP) published a report on the nation's public health newborn screening programs. This report supports the CORN guidelines to establish effective newborn screening systems, and emphasized the need to ensure newborn screening programs are adequately funded and fully integrated with the health care delivery system. The AAP identified that an effective newborn screening system should include smooth integration of sample collection, laboratory testing, follow-up, diagnosis, timely treatment, and tracking components (*Serving n. pag.*).[49] Advances in technology and the national impetus to adequately serve children with special health care needs that are identified through screening provide new challenges for public health to implement screening programs that are effective, cost-efficient, available to all infants regardless of ability to pay, and provide services beyond the newborn period into adulthood.

The Five Parts of a Newborn Screening System

1. Screening: Testing of newborns.
2. Follow-up: Rapid location, follow-up, and referral of the screen-positive infant.
3. Diagnosis: Evaluation of the infant with a positive screening test to make a definitive diagnosis or exclude the disorder.
4. Management: Rapid planning and implementation of long-term therapy.
5. Evaluation: Validation of testing procedures, assessment of the efficiency of follow-up and intervention, and assessment of the benefit to the patient, family, and society.

(Pass et. al. 3)[44]

The following (pages 49 to 56) provide an overview of the Oklahoma Newborn Screening Program's issues and activities, and family stories.

[49] *Serving the Family…*

[44] *US Newborn Screening...*