Association of Public Health Laboratories Statement prepared for the Secretary's Advisory Committee on Heritable Disorders and Genetic Diseases in Newborns and Children Secretary of Health and Human Services September 22-23, 2004

The Association of Public Health Laboratories (APHL) represents U.S. public health laboratories in all 50 states and six territories, linking them with federal partners, such as the Centers for Disease Control and Prevention (CDC) and Health Resources and Services Administration (HRSA), as well as county, local, and international laboratories. Public health laboratories have been responsible for newborn screening since the mid-1960's and currently conduct approximately 97% of all newborn screening tests. More than four million newborns are screened each year in the U.S. for inherited conditions and other disorders. Scientific studies have proven our nation's comprehensive newborn screening system of quality testing, rapid reporting, diagnosis, treatment, and care prevents death, mental retardation, and other serious disabilities.

All current public US newborn screening programs operate through the auspices of a state public health department. In most states, the state public health laboratory performs the testing, while in others, the actual laboratory testing is performed by a contract laboratory, which may be another state laboratory or private laboratory. If one state public health laboratory is down, due to hurricane, fire, bioterrorism or other natural or man-made disaster, the specimens can be directed to another state. APHL connects the state public health laboratories conducting newborn screening, allowing for more effective Continuity of Government (COG) and Continuity of Operation (COOP), a critical role for the laboratories and the association.

APHL has long provided leadership in the development and delivery of newborn screening systems. APHL and CDC co-sponsor the Newborn Screening Quality Assurance Program (NSQAP), which assesses the quality of testing done by laboratories charged with detecting inborn errors of metabolism and other disorders. Every state's newborn screening laboratory participates in this proficiency testing program. NSQAP is a voluntary, non-regulatory program to help state health departments and their laboratories maintain and enhance the quality of test results. The program provides services to more than 73 domestic newborn screening laboratories, 28 manufacturers of diagnostic products, and 387 laboratories in 53 countries. For over 26 years, NSQAP has been providing comprehensive quality assurance services for dried-blood-spot testing. The NSQAP helps state newborn screening laboratories to produce the highest possible quality results.

APHL's Newborn Screening and Genetics program strengthens the role of public health laboratories in genetics testing and designs strategies to address changes in the newborn screening testing field. This program has been funded over the past three years by the National Center for Environmental Health, Centers for Disease Control and Prevention. The Newborn Screening and Genetics in Public Health committee at APHL is charged with coordinating and promoting newborn screening projects and symposia as well as developing and adopting position statements on newborn screening and genetics. APHL's Quality Assurance/Quality Control/ Proficiency Testing Subcommittee guides the organization on newborn screening quality assurance issues.

All of the issues under discussion today affect APHL and our members, the state public health laboratories. The recent boon of technology and increasing knowledge of genetics has highlighted a growing need to incorporate advanced testing technologies into public health laboratories, as well as to explore the public health laboratory's role in testing for genetic conditions. APHL is responsible for training state public health laboratory technologists and follow up coordinators on the interpretation of results from new technologies such as tandem mass spectrometry. APHL, CDC, HRSA and the National Newborn Screening and Genetics Resource Center co-sponsor two types of training workshops on tandem mass spectrometry; one dealing with the technical application of MS/MS and the other on the challenges of interpretation and

follow up. To date, APHL has enabled the training of primary operators of tandem mass spectrometers in 25 states.

Soon your committee will address how state laboratories are funded to accomplish the important work of newborn screening. If the committee agrees to recommend a uniform panel of thirty or more disorders, APHL urges the committee to address the financial requirements of this decision. A decision to require an expanded uniform screening panel must be accompanied by expansions in equipment, personnel, training and even physical space in some instances. Capital funds for start up are required plus the expense of acquiring new and back up equipment, maintenance of equipment and kits. Adequate funding and continued extensive training for additional full time staff with expertise in MS/MS, follow up coordinators and nurses will also be crucial in sustaining newborn screening programs. None of the expenses associated with these changes are minimal, and funding increases at multiple levels of government will be necessary to achieve the desired results of the expanded panel. Screening for thirty or more disorders obligates a state to have access to highly advanced instrumentation, including tandem mass spectrometry capabilities. Today, there are not enough mass spectrometers in the country to immediately implement an expanded panel of screening. In order to implement an expanded newborn screening panel on a national basis, there must be an understanding of the funding mechanism to support it.

The mechanisms by which programs pay for the laboratory testing and follow-up services vary considerably. Funding mechanisms for newborn screening are dictated by the organizational structure of individual programs. Newborn screening financing is a state-specific infrastructure issue, and it is incumbent upon each state to provide adequate resources for the program. It is APHL's belief that resources generated by, or allocated to, a newborn screening program should be specifically earmarked for all phases of the newborn screening system (newborn blood specimen collection and transport, laboratory screening, follow-up, therapy, quality assurance, education, evaluation), program development and adaptation of new technologies. Consideration for financing the entire newborn screening system including diagnosis, medical management, and information technology should be developed in collaboration with state program directors of all relevant components as well as the health care providers and payers who will be responsible for the medical management of the identified infants.

In summary, public health laboratories currently perform a majority of newborn screening in the U.S. The impact of newborn screening is indisputable. It is a success story that demonstrates the deep commitment and quality la boratory practices that state public health laboratories bring to the newborn screening system. APHL embraces the opportunity to improve child health through fully utilizing new genetic and chemistry screening technologies. APHL will continue to provide guidance to states on expanded screening including advocacy for federal dollars to support expansion and training to assure quality programs and strongly believes federal financial support will be necessary to assure that children in all states benefit.

For more information on APHL please contact Jelili Ojodu at 202.822.5227 x235, jojodu@aphl.org

2025 M Street, NW Suite 550 Washington, D.C. 20036
202.822.5227 FAX 202.887.5098 website: www.aphl.org