

New Technologies in Newborn Screening: A View from the States

The Association of Public Health Laboratories (APHL) and the Council of Regional Networks for Genetic Services (CORN) are encouraged about new testing technologies, such as DNA analysis and tandem mass spectroscopy. There is an evolving consensus that these technologies can significantly augment newborn screening programs, if implemented intelligently and in the context of a system which includes rapid retrieval, referral, diagnosis, management and system evaluation.

Newborn screening is more than just a laboratory activity; it is an essential preventive public health program. As such, it must assure universal screening, follow-up, treatment and long term management of detected newborns. The system includes nurses, physicians, laboratorians, and other health care providers. All participants in this system are committed to embracing advances in newborn screening laboratory science in order to improve the methods for detecting, characterizing, and treating infants born with metabolic or genetic disorders.

Over the past 35 years, state and territorial governments, with support from various agencies such as the Department of Health and Human Services, have established programs with the necessary knowledge and infrastructure to detect and prevent severe mental retardation and sudden death in affected newborns. These programs have significantly reduced or eliminated the perinatal, postnatal and long term complications of infants with PKU, hypothyroidism, sickle cell disease, galactosemia and several other congenital disorders.

States and territories have judiciously committed limited resources to newborn screening programs when it was clear that screening would be efficiently and effectively conducted for diseases which were well characterized, had significant prevalence in the population being screened, and were treatable with cost-effective therapies administered in a timely manner. State and territorial health departments have the responsibility to expend limited resources to the greatest advantage of the population. This is a time of increasing emphasis on cost containment, cost effectiveness, and rigorous evidence-based medicine in the delivery of all health care services. As additional diseases were defined, and therapies characterized, public newborn screening programs committed additional resources to screening for some of these diseases, and this continues. For example, as testing for cystic fibrosis gains support for possible addition to testing batteries, pilot projects are establishing the efficacy of screening so that appropriate funding decisions can be made.

New testing technologies, such as tandem mass spectroscopy, must be carefully evaluated by well-designed pilot studies before they are integrated into screening programs. There are many unanswered questions concerning the use of these technologies and federal funds should be made available to conduct these research studies. These evaluations should follow the guidelines developed by recognized professional groups involved in population-based screening. Judgement on their use should not be made on anecdotes and advertising by for-profit companies. New testing technologies should be used by public health-based newborn screening programs wherever possible, but within the limits of most effective and efficient use of public resources.

Consistent review and evaluation will assure that the most appropriate and cost-effective detection, follow-up and treatment protocols are implemented for the benefit of the greatest number of affected newborns.

APHL Neonatal Screening Committee
CORN Newborn Screening Committee
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