

Emerging Issues and Technologies in Newborn Screening and Genetics Initiatives

Emerging issues and technologies in newborn screening and genetics initiatives are activities of the Genetic Services Branch that help State maternal and child health (MCH) officials, health care providers, public health professionals and families, and individuals respond to new scientific findings and technologies in the fields of MCH genetics and newborn screening, with special consideration of the financial, ethical, legal, and social implications surrounding the uses of genetic medicine and technology in MCH populations.

As molecular technologies and the understanding of newborn and child congenital disorders move forward, the opportunity to screen newborns and children early for many commonly occurring congenital disorders increases. The benefits of detecting these disorders early include health promotion and disease intervention strategies.

However, before States can promote the widespread use of molecular technologies for newborn and child screening and diagnosis, many screening issues must be resolved. Some of the questions that are currently being asked include:

- Should States enhance or expand newborn and child population-based screening, and where should such screening occur?
- Which genetic disorders should be included in the newborn screening battery?
- What methods, quality controls, and regulatory and legislative measures are necessary to ensure the maximum benefits for patients and families?
- What types of parental and individual consent should be used?
- What safeguards should be in place to assure safety, privacy, and access to genetic medical information?
- What is the meaning of fairness in the distribution of the costs and benefits of newborn screening?
- How will these and future questions be resolved?

Activities

To address emerging issues that arise with State-based newborn screening and genetics programs, the Genetic Services Branch supports several initiatives that help MCH health care and public health professionals and decision-makers formulate the most appropriate practices and policies. The Branch works to address the many ethical, legal, and social issues arising from the use of new and evolving technology within newborn screening and other genetic service programs. These activities are as follows:

Genetic Services Branch Workshops have been convened each year since 1999. The purpose of these sessions is to educate State legislators and State health officials on genetics issues and newborn screening, including the National Newborn Screening Task Force's recommendations. In addition, the workshops address concerns related to the potential misuses of genetic information such as discrimination, as well as various privacy and confidentiality issues.

The **National Newborn Screening and Genetics Resource Center** offers technical assistance to States regarding the use of newborn screening technologies.

The **Departments of Health of California and Massachusetts** each have been awarded a 3-year grant in 2001 to identify models for evaluating the clinical validity and utility of the use of tandem mass spectrometry in newborn screening programs; both of these grants represent collaborations between multiple states. **State of Hawaii Department of Health** was awarded a 3-year grant in 2001 to identify new models and materials that address financial, ethical, legal, and social issues related to using new genetic screening technology in newborn screening programs. The findings of this program are expected to be of practical use by the Maternal and Child Health Bureau and newborn screening programs in various States.

The **University of California, Los Angeles (UCLA)** was awarded a 4-year contract in 2001 to examine current informed consent processes, policies for newborn blood spot screening samples, and educational materials related to newborn screening. Based on these findings, UCLA will develop educational materials, and model policies and procedures for the informed consent process and for storing, using, and accessing newborn blood spot screening samples.

The **Hastings Foundation** was awarded a contract in 2001 to examine the issue of fairness in the distribution of costs and benefits of newborn screening.

Resources

The **National Newborn Screening and Genetics Resource Center** offers newborn screening and genetics resources for the MCH health professionals, the public health community, consumers, and government policymakers. To see a complete set of offerings, visit <http://genes-r-us.uthscsa.edu> or call (512) 454-6419.

GeneTests-GeneClinics is a publicly funded medical genetics information resource developed for health care providers and researchers. To learn more, visit www.geneclinics.org.

The **National Coalition for Health Professional Education in Genetics** is a coalition of more than 100 health organizations. To learn more, visit www.nchpeg.org.

The **HRSA Information Center** offers a wealth of maternal and child health publications and resources. To see a complete list of offerings, visit www.ask.hrsa.gov or call toll-free (888) ASK-HRSA (275-4772).

MCHB Program Grants Guidance and Application Materials are available online. Those interested in applying for MCHB grants should visit www.mchb.hrsa.gov/html/grantsguidance to access instructions and other pertinent information.

The Genetic Services Branch is in the Division of Services for Children with Special Health Needs, which is part of the Maternal and Child Health Bureau. For more information, call (301) 443-2170, or visit www.mchb.hrsa.gov.