

# Newborn Screening and Genetics Public Health Infrastructure

**Newborn screening and genetics public health infrastructure initiatives** help support State newborn screening and genetics programs, integrate newborn and genetic screening programs with other community services and medical homes, and strengthen existing newborn and genetic screening and service programs.

Since most diseases are influenced by genetic risk factors, advances in genetic medicine need to be integrated into health promotion and disease prevention activities in both maternal and child health (MCH) public health and primary health care programs. Historically, genetic services have focused on health promotion, prevention, health education, and integration with primary and subspecialty care. Many States have organized family-based services, which include general genetics clinics, as well as clinics for specific genetic-based conditions, such as metabolic disorders and hemoglobinopathies or hemophilia. Other genetic services have included population-based efforts within newborn screening programs and maternal screening programs. Many States have organized specific laboratory services for cytogenetics, newborn screening, molecular genetics, maternal screening, and biochemical genetics.

Advances in science and technology have led to the development of many different types of newborn screenings necessitating an evaluation of newborn screening programs. In August 2000, a task force convened by the American Academy of Pediatrics and the Health Resources and Services Administration' (HRSA) Maternal and Child Health Bureau (MCHB) released a report on newborn screening. It outlined MCHB's national agenda for strengthening State newborn screening programs. Guided by this agenda, the Genetic Services Branch is making great strides toward improving coordination and uniformity among State newborn screening programs across the country.

## Activities

All States and the District of Columbia have newborn screening programs. These programs vary in the number and types of medical conditions for which they test. Currently, all States screen for phenylketonuria (PKU), which causes mental retardation unless children begin a special diet immediately after birth, and hypothyroidism, an easily treated hormone deficiency. Newborn screening and genetics public health infrastructure initiatives seek to build on these programs to implement projects in the following areas:

- Needs assessment for State newborn screening programs, genetics programs, and service systems
- Integration of newborn screening and genetic services for newborns and children who have, or are at risk of having, heritable disorders into existing State systems of care, networks of services and supports that meet the diverse and changing needs of mothers, children, and families, including those with special health needs
- Evaluation of new technologies, such as the use of tandem mass spectrometry and DNA-based technologies for diagnosis and in screening programs

- Development of guidelines for genetic and newborn screening services and uniform State newborn screening program standards

Another important newborn screening and public health infrastructure initiative is the **National Newborn Screening and Genetics Resource Center**. This center provides newborn screening and genetics resources that benefit families, MCH health professionals, the MCH public health community, and policymakers. The Center also helps States improve newborn screening and genetic service activities through onsite technical reviews.

## Resources

The **National Newborn Screening and Genetics Resource Center** offers newborn screening and genetics resources for MCH health professionals, the public health community, families, 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](http://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](http://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](http://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](http://www.mchb.hrsa.gov/html/grantsguidance) to access instructions and other pertinent information.

*Newborn screening and genetics public health infrastructure initiatives are administered by the Genetic Services Branch, 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](http://www.mchb.hrsa.gov).*