Newborn screening is a public health success story, enabling the early identification of infants with potentially life-threatening heritable disorders and genetic diseases. Roughly four million infants are screened for disorders each year in the United States, however there is little data illuminating the health outcomes of the thousands of infants who are ultimately diagnosed with a newborn screening condition each year. Compared to the process of screening and diagnosis, fewer resources have been devoted to long-term follow-up (LTFU) activities that are necessary to limit the health consequences of confirmed disorders and to document and strengthen the performance of newborn screening systems.

Maternal and child health advocates recognize that LTFU activities are essential to achieve the full promise of newborn screening. This report outlines critical components of LTFU for policy makers and administrators of state newborn screening programs and opportunities to help states achieve comprehensive LTFU systems.

A Framework: Components of LTFU

Drawing on work from the American College of Medical Genetics,1 the Clinical and Laboratory Standards Institute,2 and the National Newborn Screening and Genetics Resource Center,3 the Association of Maternal and Child Health Programs (AMCHP) proposes that the basic components of LTFU are:

- Assuring that ongoing, high-quality medical management—including specialty care and care coordination, when needed—are provided within the context of a medical home to children, adults and families affected by heritable disorders;
- Assuring transition of young adults to appropriate adult medical care;
- Periodically assessing patient progress through review of defined outcome indicators;
- Collecting and analyzing state LTFU data; and,
- Engaging in continuous quality improvement at the local, state and national levels.

LTFU begins after short-term follow-up ends; that is, with the initiation of treatment. For the purposes of maternal and child health (MCH) program responsibility, LTFU ends upon transition to the adult service delivery system at 21 years of age. Concurrent with such a transition, public health agencies should assure the formal transfer of LTFU responsibility from MCH to an appropriate public health program to continue LTFU throughout the lifespan of affected individuals.

Current State LTFU Activities

The AMCHP Newborn Screening and Genetics Advisory Group surveyed a sample of state newborn screening and MCH programs to determine existing state LTFU activities. The group found that:

- Even in states assumed to be most actively pursuing LTFU activities, the activities are limited and generally have not progressed beyond early stages of development;
- Typically, LTFU activities are divided among several state programs, with the specific division of responsibilities varying from state to state;
- States are most actively engaged in the first component of LTFU listed above: assuring that high-quality medical management and care coordination are provided to children, adults and families affected by heritable disorders;
- The level of clinical tracking varies greatly for each disorder category; and,
- States have experimented with innovative approaches to LTFU, particularly when dedicated federal funding has been available.

Challenges and Opportunities

The biggest challenges facing those who wish to strengthen LTFU systems are the low priority accorded to LTFU compared to other newborn screening activities and the consequent scarcity of resources. However, newborn screening advocates can confront these challenges by capitalizing on several opportunities:

Timeliness: The April 2005 release of the first guidelines from the American College of Medical Genetics to define a uniform panel of conditions for state-mandated newborn screening, provides a compelling need and critical opening to examine the activities required to support an expanded testing panel and associated increase in the number of infants diagnosed with rare disorders.

Ongoing Projects: Federally supported projects have instigated nascent efforts to develop LTFU datasets, medical home pilot projects and other initiatives. As these efforts are further developed, projects can share sample datasets, model policies and other products for adaptation by others.
Uniform Core Datasets: Promotion of uniform core datasets would be cost effective, increase the data’s statistical power—especially for rare disorders—and facilitate continued LTFU as patients and their families move within and between states.

Building on Existing Data Systems: Existing vital registration databases, birth defects registries, immunization registries, newborn screening short-term follow-up files and genetics databases can be integrated with newborn screening data systems to promote efficiency and collaboration across health programs, facilitate research, and ultimately enhance continuity-of-care for children and families.

Next Steps

The AMCHP Newborn Screening and Genetics Advisory Group recommends that states focus on collaborative integrated planning efforts such as those initiated by the Genetic Service and Newborn Screening Regional Collaborative Groups. The recommendations listed below would help to further develop newborn screening LTFU systems.

1. Publicize and advocate that LTFU is an essential part of a quality newborn screening program and a public health responsibility;
2. Develop an implementation plan for the LTFU components outlined in this report;
3. Secure funding for LTFU;
4. Develop standards for data collection related to newborn screening LTFU; and,
5. Provide resources and technical assistance to states as they develop LTFU activities.

Endnotes

1 American College of Medical Genetics, Newborn Screening Expert Group. Newborn Screening: Toward a uniform screening panel and system. Genetics in Medicine. 2006; 8(S) Supplement: 12S-252S.

2 Newborn Screening Follow-up; Approved Guideline. CLSI document I/LA27-A, 26(18), 2006.

3 National Newborn Screening and Genetics Resource Center. Program Evaluation and Assessment Scheme (PEAS). Posted at genes-r-us.uthscsa.edu/NBS_PEAS.htm.

This report was prepared by AMCHP’s Newborn Screening and Genetics Advisory Group. For a full copy of the report and more information about the key resources and partners highlighted in this summary, please refer to the AMCHP website: www.amchp.org.