



Advisory Committee on Heritable Disorders and Genetic Diseases in Newborns and Children

June, 2006

Health Resources And Services Administration
Maternal And Child Health Bureau

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Maternal and Child Health Bureau

Mission: To provide national leadership and to work, in partnership with states, communities, public-private partners, and families to strengthen the MCH infrastructure, assure the availability and use of medical homes, and build the knowledge and human resources, in order to assure continued improvement in the health, safety and well-being of the MCH population.



Maternal and Child Health Bureau

The MCH population includes all America's women, infants, children, adolescents and their families. Includes individuals across their life span, women of reproductive age, fathers, and children with special health care needs.



Maternal and Child Health Bureau

Current Newborn and Genetic Service Activities



Present Initiatives

- Education
- Training
- Capacity
 - Infrastructure
 - Workforce
- Research
- Public Policy



MCHB's Vision for Newborn Screening

- Systems approach with defined public health roles at state and national level
- Presence of quality assurance
- Public–private partnerships for assurance of systems approach and comprehensive, efficient care and management
- Equity for families



Resource Center

National Newborn Screening and Genetics Resource Center

<http://genes-r-us@uthscsa.edu>

Serves as a focal point for national newborn screening and genetics activities, and provides related resources to benefit consumers, health professionals, the public health community, and government officials.

- Newborn screening expert consultation to state and territorial public health agencies
- Internet-based, real-time national newborn screening information retrieval system (NNSIS)
- MS/MS training workshops to assist state NBS programs (co-sponsors CDC and APHL)



Genetic Services and Health Care Delivery

Support the Hemophilia Diagnostic and Treatment Centers and Thalassemia and Sickle Cell Disease programs as **models of comprehensive care** for the delivery of genetic services: testing, counseling, education and coordinated system of services.



Programs

- **Hemophilia Program**-Provide access to hemophilia diagnostic and treatment centers to provide comprehensive care. Program also provides access to pharmaceuticals at a discounted price (340 B program) and passes the savings on to the consumers or back into services.
- **Sickle Cell and Newborn Screening**- Support models of delivering SCD and SCD carrier counseling and follow-up and linkage to newborn screening and comprehensive system of care.
- **Sickle Cell Disease Treatment Program (NEW)**-Develop and establish mechanisms to enhance the prevention and treatment of sickle cell disease through the coordination of service delivery; genetic counseling and testing; bundling of technical services; training of health professionals; and other related efforts.
- **Thalassemia**- Support implementation of demonstration grants regarding a model system of comprehensive care and medical management for individuals and families at risk for or affected by Cooley's Anemia/Thalassemia-tertiary care.

Understanding, Informing and Educating Parents About Newborn Screening

Analysis of:

- State Statutes/Regulations/policies regarding consent for newborn screening including recommendations for a state resource tool kit.
- State Statutes/Regulations/policies regarding storage and use of residual blood spots following newborn screening including recommendations for a state resource tool kit.
 - Published in supplement to PEDIATRICS (June)
 - Conducted in collaboration with the National Conference of State Legislatures



Understanding, Informing and Educating Parents About Newborn Screening

Develop educational materials for prenatal providers for educating parents

- Partnership with AAP, ACOG, AAFP
- Target: health professionals with the primary responsibility for prenatal health care, labor and delivery services (obstetricians, family practice physicians and nurse midwives)
- **7 Things Parents Want to Know about Newborn Screening**
- **Quick Reference Guide for Health Professionals**
- Adopted by AAP and ACOG
- Published in supplement to PEDIATRICS (June)

Understanding, Informing and Educating Parents About Newborn Screening

7 Things Parents Want to Know About Newborn Screening:



THE HEALTH PROFESSIONAL'S GUIDE FOR BRIEF DISCUSSION WITH PARENTS.

1. All newborn babies are required by the State to get tested for some rare disorders before they leave the hospital.
2. Babies with these disorders may look healthy at birth.
3. Serious problems can be prevented if we find out about the disorders right away.
4. To do the test, a nurse will take a few drops of blood from your baby's heel.
5. Your baby's doctor and the hospital will get a copy of the test results. Ask about the results when you see your baby's doctor.
6. Some babies will need to be retested. If your baby needs to be retested, you will be notified. It is very important to get retested quickly.
7. Talk to your baby's doctor if you have questions. The Web site on the back of the brochure also has good information.



U.S. Department of Health and Human Services
HRSA
Health Resources and Services Administration



Understanding, Informing and Educating Parents About Newborn Screening

- Develop educational sample templates for State NBS programs and healthcare providers for educating parents:
 - These Tests Could Save Your Baby's Life – answers basic questions about newborn screening

THIS TEST COULD SAVE YOUR BABY'S LIFE



Why does my baby need Newborn Screening tests?

Most babies are just fine when they are born.

We test all babies because a few babies look healthy but have a rare health problem.

If we find problems early, we can help prevent serious problems like mental retardation or death.

How will my baby be tested?

Before you leave the hospital, a nurse will take a few drops of blood from your baby's heel.

The hospital will send the blood sample to a newborn screening lab.

How will I get the results of the test?

Parents are notified of test results if there is a problem.

Ask about results when you see your baby's doctor.



Why do some babies need to be retested?

All babies who leave the hospital early must be retested.

Some states require a second test on all babies.

Some babies need to be retested because there is a problem with the blood sample.

A few babies need to be retested because the first test showed a possible health problem.

What if my baby needs to be retested?

Your baby's doctor or the State Health Department will contact you if your baby needs to be retested. They will tell you why the baby needs to be retested and what to do next.

If your baby needs to be retested, get it done right away.

Make sure that your hospital and doctor have your correct address and phone number.

What if I have questions?

Ask your baby's doctor if you have questions or concerns.



Brochure has been produced & distributed by AAP and ACOG. An electronic template is available for states to use or modify.



Heritable Disorders Program

Regional Genetic Service and Newborn Screening Collaboratives

- Enhance and support the genetics and newborn screening capacity of States within defined regions
- These projects will undertake a regional approach toward addressing the maldistribution of genetic resources
- 7 regions and a national coordinating center



Other Projects

Consumer based family history tool to increase the public's awareness of genetics

- Partners: Library of Congress, Genetic Alliance, American Society of Human Genetics
- Toolkit to be piloted over the next 3 years

Translational Genetic Services

- Analysis of models of genetic service delivery, including economic and policy issues;
- Discussion, dialogue and agenda setting to address the translation of genetic research into practice;
- Cooperative Agreement with Washington State



Other Projects (continued)

Two Newborn Screening projects:

1. Establish a quality assessment and evaluation scheme for newborn screening programs (NNSGRC)
 - Assistance to develop indicators for system components
 - Evaluation by selected programs to be conducted
2. Support a newborn screening informatics practice network to develop best practices for newborn screening program and data integration projects
 - Public Health Informatics Institute serves as a technical assistance and resource center



Other Projects (continued)

- 1. ACT(ion) sheets and confirmatory algorithms**
 - Developed through ACMG for conditions included in newborn screening panels. Many States are adapting the materials for their programs for distribution to pediatric health professionals with newborn screening results
 - On ACMG website
<http://www.acmg.net/resources/policies/ACT/condition-analyte-links.htm>.
 - Adopted by AAP Board. Conduct survey through AAP on utility
- 2. Supplement to PEDATRICS**
 - Summary of work since AAP Task Force Report
 - Includes cost analysis by Aaron Carroll and Stephen Downs

Contact Information

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Data Site

<https://performance.hrsa.gov/mchb/mchreports>