A New Era in Newborn Screening
Saving Lives, Improving Outcomes

CDC Satellite Broadcast – September 19, 2002
http://www.phppo.cdc.gov/PHTN/Newborn

The following questions were phoned or faxed in during the live broadcast.
There was not enough time to answer all during the program, but
all submitted questions and the answers are listed below.
Some of the live responses in this list have been condensed or expanded upon.

Questions about funding for newborn screening services:

Are funds made available to local genetic disorder support groups?

(Dr. Puryear) There are many funding resources available to community genetic disorder support groups through the federal and state governments, foundations, and certain industries. Information on federal grant opportunities is listed in the Federal Register. State departments of health may also have information on funding opportunities for genetic support groups. The Genetic Alliance, a lay advocacy coalition representing consumer and health professional organizations may also provide information on strategic ways to obtain funds.

Are newborn screening services offered to babies adopted from overseas? If so, till what age are these services available?

(Dr. Puryear) The attending health care provider for the adopted newborn should perform a newborn screening test if it had not been obtained at birth. Screening tests should be done soon after birth. Older children and adults also can be screened. Contact the child's health care provider for more information.

Will grant funds be made available to states under Title XXVI for newborn screening projects?

(Dr. Puryear) Yes, there is language in Title XXVI of the Children's Health Act of 2000 (Section 1109 of the Public Health Service act) authorizing the Secretary to award grants to states, or a political subdivision of a state, or a consortium of two or more states, or political subdivision of states to enhance, improve or expand the ability of states and local public health agencies to provide screening, counseling or health care services to newborns and children having or at risk for heritable disorders.

Who pays for the treatment and follow-up of infants diagnosed positive for any of the newborn disorder screened? Are parents responsible for the costs of special diets and medication?

(Dr. Fernhoff) Each state or region is different. To find out what is available in your state, contact your state genetic coordinator. Some states do cover the costs for special treatment and follow up. Some states do cover the costs for special formulas and foods for low protein diets, at least through the childhood period. Other states have relatively limited resources and these are not covered. Health insurance benefits vary similarly. Parents have been the advocates for change in state and insurance company policies toward assuring that these needs are met. Progress is being made although it is still an uphill battle.

Do parents have access to a broader range of screening tests for their newborn than may be offered by their state? Who can parents contact to get such information?

(Dr. Fernhoff) Different disorders are screened for among the states. Supplemental testing for those test not offered in a particular state can be performed through several private laboratories. It is always best to discuss testing issues with your baby's physician or primary care provider. In the program materials, http://www.phppo.cdc.gov/dls/genetics/nbs_091902.asp, several on-line resources have been listed that can direct you to these laboratories

The ability to treat an infant for a disorder detected by newborn screening is one of the criteria used for deciding whether a particular disorder should be included on the screening panel. How is "treatment" defined?

(Dr. Puryear) There are efforts underway at the national level to develop a set of newborn screening recommendations that are based on current scientific evidence. These recommendations are foreseen as being useful to the states to develop policies that are in the best interest of their respective populations. Each state has the responsibility for implementing and evaluating their newborn screening program. Defining "treatment" is an important discussion and involves both public health and clinical care providers and parents. Presently, there are different definitions. Generally speaking, it would imply an available therapy that would reduce the adverse symptoms in an untreated child. Questions about what constitutes "treatment" have come up especially with regards to conditions diagnosed by tandem mass spectrometry and molecular testing.

What efforts are being made to educate health care providers and the public about newborn screening?

(Dr. Puryear) There are several national projects underway designed to enhance the education of health care providers and the public. Senators Dodd and DeWine have recently sponsored a bill that if funded, will provide education for health care workers laboratory professionals, and the public. The March of Dimes also has educational materials on newborn screening, as do each of the State programs. In addition, the Health Resources and Services Administration (Maternal and Child Health Bureau, Division of Services for Children with Special Health Needs, Genetics Services Branch) have undertaken multiple public and provider education projects over the past 25 years. Projects include:

> The GENE project engages underserved communities in developing a process that will enable them to use genetic information to make more informed choices about their health. The project is also designed to improve these communities' access to culturally and linguistically appropriate genetics information, resources, and services.
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> Genetics in Primary Care (GPC): A Faculty Development Initiative to enhance the ability of faculty to incorporate the clinical application of genetic information into undergraduate and graduate primary care medical education. There are GPC sites at 20 medical schools across the country.
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> Genetics Through the Primary Care Lens - Genetics 101 project will develop a curriculum for primary care physician providers. The content of this curriculum will reflect the needs of primary care providers identified through the Genetics in Primary Care training sessions.
>
> The Centers for Disease Control and Prevention have also developed and interactive multimedia training program "Genetics in Clinical Practice: A Team Approach" to educate

primary care physicians and others about the complexities encountered addressing genetic issues with patients.

Questions about newborn screening services:

What is the process that determines which tests are offered by a particular state?

(Dr. Levy) Processes vary among states and mechanisms can include legislative mandate, directive of the state health commissioner, recommendation of an advisory panel, or action by the screening program. There are efforts to normalize this process on a national level. To find out about a particular state, contact that state's newborn screening coordinator.

Who can be contacted to obtain information about a particular state's newborn screening program?

(Dr. Puryear) Contact the newborn screening advisory committee or newborn screening program coordinator in your respective state.

You mentioned that states are either testing for MCADD or gearing up to do so. Which states presently screen for MCADD?

(Dr. Puryear) This information is available on the website for the National Newborn Screening and Genetics Resource Center: http://genes-r-us.uthscsa.edu - go to Quick Links and then access "NBS Disorders Screened -Listed by State". States screening for Medium-Chain Acyl-CoA Dehydrogenase Deficiency (MCADD) are listed as performing MS/MS.

Who can be contacted to identify persons willing to speak about newborn screening at community meetings?

(Dr. Puryear) Your state newborn screening program, state genetics coordinator, lnewborn screening advisory committee, local or state health department, or local chapter of the American Association of Pediatricians can all provide suggestions for speakers.

If a parent objects to testing for religious or other reasons, will the baby still be tested?

(Dr. Levy) Some, but not all states, allow for withdrawal from the testing process for religious and other reasons. This is a subject still being discussed in which consensus has not been reached.

If blood spots are sent out of state for testing, who determines which tests are performed on the blood spots?

(Dr. Levy) The state where the sample originated determines which tests are performed. Only laboratories that can perform these tests will be used. For instance, in New England, there are five states that send their samples to a laboratory in Massachusetts. Each state requires a different panel to be performed. The laboratory is able to perform the requested set of tests from each of the states that sends samples.

Questions about newborn screening tests and treatments:

If a screening test comes back abnormal does it mean that my child is going to be really sick?

(Ms. Quary) It is important to recognize that screening tests do not diagnose the presence of a disease but identifies those infants who are at higher risk for having a disease. Follow up diagnostic testing is necessary to determine if a disease is actually present. False positive results do occur with screening tests and so follow up with one's physician for diagnostic testing is necessary. Newborn screening identifies persons who should be referred to diagnostic testing so an appropriate and timely clinical decision can be made.

Should a blood specimen be obtained from an infant prior to an emergency transfusion, even if the baby is not yet 24 hours old?

(Dr. Pass) Yes, a blood specimen should be obtained before transfusion. Different tests have different requirements. Those that measure some property of red cell enzymes or red cell components certainly would be more valid prior to transfusion than other tests. In addition to taking a sample prior to the transfusion, another sample should be collected at about 24 hours post-transfusion to allow the serum to equilibrate. A third sample should be taken about 30 days post-transfusion. Analysis of this third sample helps the lab screen for problems with red cell components.

What recommendations are available or efforts underway to address the issue of newborn samples submitted from babies that have been transfused? Specifically, are there recommendations with regard to testing T4 and TSH levels for hypothyroidism?

(Dr. Levy) The recommendations concerning specimen collection from transfused babies are general and do not specify particular disorders. Nevertheless, they should apply to all of the screened disorders. In Massachusetts, for example, the recommendation is that a specimen should be collected before the transfusion regardless of the age of the baby and a repeat specimen collected at least 2 days after the transfusion. Finally, another specimen should be collected at 2 months after the baby's final transfusion.

Are the results obtained from screening a pre-term baby as reliable as one full-term?

(Dr. Levy) Generally yes. However, there is concern about the possibility of missing a premature baby with congenital hypothyroidism because of delay in the rise of TSH. Consequently, at least some states recommend collection of a second specimen at two weeks of age or at nursery discharge, whichever is earlier, from premature babies. More likely is the problem of false positives (lack of specificity) in premature infants, especially very small premies. These infants frequently have transient increases that require repeat testing to rule out a disorder.

If the father has PKU, what affect will this have on the unborn child and how does this differ if the mother has PKU?

(Dr. Levy) None whatsoever, in contrast to a mother having PKU.

Will the test panel continue to expand and what's next?

(Dr. Pass) Yes. The panel will expand. Efforts are underway to develop screening programs for other diseases including such chronic conditions as asthma and diabetes. The full

potential of tandem mass spectrometry has not yet been realized. The blood spot sample is no longer a limiting factor since analyses can be performed on minute quantities. Another new area is molecular (DNA-based) testing which offers significant opportunities for additional testing.

How can I find out more about the laboratory diagnosis of Sickle Cell and some of the other diseases?

(Ms. Quary, Dr. Puryear) The best course is to contact your state newborn screening laboratory or follow-up program or local health department. In the program materials, http://www.phppo.cdc.gov/dls/genetics/nbs_091902.asp, the "Related On-line Resources" contains additional information including The Sickle Cell Information Center, National Newborn Screening and Genetics Resource Center and parent support groups.

Is the PKU diet required throughout life or until age 6?

(Dr. Puryear) Dietary discontinuance before 8 years of age is associated with poorer performance on measures of IQ. The effects of dietary discontinuance at older ages (12 years and above) are less clear. Adults with PKU who are not on restricted diets show stable IQ scores, but also manifest poorer performance on measures of attention and speed of processing. Higher levels of phenylalanine are accepted with frequent monitoring. Evidence shows that the individual with PKU must be maintained on a lifelong restricted phenylalanine diet, though some relaxation may be tolerable, in some cases, as the individual ages.