

**Secretary's Advisory Committee on Heritable Disorders
and Genetic Diseases in Newborns and Children**

**Summary of Third Meeting
January 13-14, 2005
Washington, DC**

The Secretary's Advisory Committee on Heritable Disorders and Genetic Diseases in Newborns and Children was convened for its third meeting at 9:00 a.m., Thursday, January 13, 2005, in the Rotunda Ballroom, Ronald Reagan Building and International Trade Center, Washington, D.C. The meeting was adjourned at 2:20 p.m., Friday, January 14, 2005. In accordance with the provisions of Public Law 92-463, the meeting was open for public comments from 1 p.m. to 2:00 p.m. on Friday, January 14, 2005.

January 13, 2005

HHS Welcome and Opening Remarks

Dennis Williams, Ph.D., M.A.
Deputy Administrator
Health Resources and Services Administration (HRSA)

Dr. Williams welcomed members of the Advisory Committee on Heritable Disorders and Genetic Diseases in Newborns and Children (the Committee) and conveyed the greetings of Secretary of Health and Human Services (HHS) Tommy Thompson and HRSA Administrator Elizabeth Duke. He noted that HHS Secretary Tommy Thompson would soon leave his post and former Utah Governor Mike Leavitt would assume the leadership. Dr. Williams commended Secretary Thompson's leadership on issues that relate to the welfare of the nation's mothers and children and thanked Dr. Howell and Dr. Peter van Dyck, HRSA Associate Administrator for Maternal and Child Health (MCHB), for their leadership on the Committee. In addition, he welcomed the newest member of the Committee, Dr. Joseph Telfair.

Dr. Williams emphasized that HHS and HRSA value the work of the Committee to assure that all children receive quality care in all States. The Committee will evaluate many critical issues surrounding newborn screening programs including: opportunities for research; information systems to evaluate long-term health outcomes of infants identified; surveillance systems to evaluate newborn screening programs; the Regional Collaboratives established this past year by HRSA's MCHB; financing of newborn screening programs; and new technologies, such as tandem mass spectrometry (MS/MS). Dr. Williams and others at HRSA look forward to the Committee's recommendations on how to improve newborn screening services nationwide and are very confident that the Committee's knowledge and expertise will help them recommend guidelines to Secretary Thompson's successor that will greatly benefit children and their families across the United States.

Committee Business

R. Rodney Howell, M.D.

Chair, Advisory Committee on Heritable Disorders and Genetic Diseases in Newborns and Children (ACHDGDNC)

Dr. Howell thanked Dr. Williams and the leadership at MCHB for their support provided to the Committee and his appreciation for the work of Committee members.. The first item of business was the unanimous approval of the minutes from the 2nd meeting of the Committee, September 22-23, 2004. Next, Dr. Howell reviewed the agenda for the 2-day meeting, which focused on Newborn Screening Program Infrastructure and will include presentations on Data Management Programs and the HRSA funded Regional Genetics and Newborn Screening Collaboratives. Recommendations from the report produced by the American College of Medical Genetics for HRSA were also to be discussed.

Dr. Howell introduced, the new Committee member, Dr. Joseph Telfair, representing the Secretary's Advisory Committee on Genetics, Health, and Society.

State of the States Update

Bradford Therrell, Ph.D.

Director

National Newborn Screening and Genetics Resource Center

Dr. Therrell gave an update on newborn screening activities in the States. He referred to slides that showed the expansion of screening panels due to the implementation of MS/MS technology. Dr. Therrell noted that the Tables of data presented are only as comprehensive as how extensively States agree to voluntarily report information. He urged the Committee to further discuss the issue of mandated reporting.

Committee members posed a number of questions. Dr. Boyle asked whether most States are required to report on newborn screening to their State health departments, and submit aggregate data to the Federal level. Dr. Therrell responded that Title V of the Social Security Act (Maternal and Child Health Block Grant) requires reporting for some disorders. State newborn screening programs may develop data and report it for their own quality control programs. This inconsistency makes it difficult to keep up with the status of State programs. There is no requirement for the States currently to report conditions to any group.

Discussion indicated that disorders are not counted consistently in the same way across States. Dr. Therrell indicated the impetus to use higher numbers is driven, in part, by competition for control of the newborn screening program or for direct marketing to parents. With the competitiveness among States and among private laboratories for how many conditions are tested, it would be helpful for the Committee to arrive at some generally agreed upon way to count conditions. Dr. van Dyck and a small group are going to produce a piece on this issue for the Committee before the next meeting to spur further thought on this issue. Dr. Howse said the March of Dimes believes that the list of tests in the ACMG report is adequate and a good starting point.

Dr. Therrell led the next discussion on the major incentives for expanded screening. In the next two to three years, consumer and professional interest and advocacy will continue through the press, legislature, and the health departments. Dr. Becker commented Ohio experienced pressures to expand its newborn screening program that would include disorders that can be screened using MS/MS technology. Because Ohio initially focused its attention on MS/MS disorders, the State is now adding cystic fibrosis, biotinidase deficiency and congenital adrenal

hyperplasia (CAH), which require technology that is not MS/MS driven. Dr. Rinaldo observed that the main driver of expanded screening is States feel they are behind, and their constituencies realize their children or babies are not being offered a perceived national standard of care. Dr. Rinaldo then suggested, the issue of counting disorders is a bit exaggerated—and the serious consequence of continued thinking along these lines is to hijack the concept of quality.

Dr. Telfair inquired about the extent of State data collected on the outcome of infants who test positive and the effectiveness of follow-up. Dr. Therrell replied, follow-up is an issue, as some States do short-term follow-up adequately and report that information; but long-term follow-up is not tracked as completely.

The topic of subcommittee structure concluded the discussion. Dr. Boyle asked Dr. Therrell to provide some guidance to the Committee on subcommittee structure, such as pressing issues and how the Committee can be most helpful in that regard. In addition, Dr. van Dyck stated that he will present a priority list of issues related to newborn screening, developed by staff that might feed into the deliberations about subcommittees.

Newborn Screening Program Infrastructure; Research and Service Delivery

Research - The NIH Office of Rare Diseases: Opportunities for Collaborative Research and Development Activities

Stephen Groft, Ph.D.
Director
Office of Rare Diseases
National Institutes of Health

Dr. Groft presented an overview of the activities of the NIH Office of Rare Diseases (ORD), including its newly established Rare Diseases Clinical Research Network and Data and Technology Coordinating Center. ORD was established in 1993 without any legislative mandate. With the assistance of patient advocacy groups, in 2002 legislation was passed that mandated certain activities within the office; the emphasis was clinical research. With this mandate came an increase in ORD's budget; the office is currently funded at the level of \$15.5 million.

The Rare Diseases Act of 2002 (Public Law 107-280) established ORD statutorily within the Office of the Director of NIH and specified the following responsibilities for ORD: (1) recommend research and public education agendas at NIH; (2) promote coordination and collaboration of rare diseases activities; (3) develop an information center on rare diseases; (4) prepare a biennial report to Congress on rare diseases research and education activities, with an emphasis on future research activities; and (5) prepare an annual report to Congress on research advances in rare diseases. ORD's biennial report is available online at <http://rarediseases.info.nih.gov>.

ORD works with NIH institutes and centers to support: (1) an extramural research program, a Rare Diseases Clinical Research Network; (2) an intramural research program, the ORD/National Human Genome Research Institute; (3) a scientific conferences program; (4) a Trans-NIH Working Group on Rare Diseases Research; and (5) a Genetic and Rare Diseases Information Center.

Committee members applauded the development of ORD's Rare Diseases Clinical Research Network. It was suggested that ORD establish links with traditional newborn screening programs

in its request for applications to obtain a repository of samples (e.g., DNA, serum, tissues) that could be utilized for its clinical investigators, and vice versa.

Dr. Boyle concluded that the Rare Disease Clinical Research Network provides an opportunity to learn more about the natural history and impact of clinical treatment on many disorders that are in the purview for newborn screening. Dr. Boyle further indicated that it appears that additional infrastructure is not needed in the area of research. Dr. Graft noted that a lot of the infrastructure already exists, but the challenge is to use it appropriately. ORD is already working closely with the NIH Institutes and with the Genetics Services Branch of the MCHB, HRSA to interconnect and share infrastructure.

Service Delivery –Infrastructure: Regional Genetics and Newborn Screening Collaboratives

Michele Lloyd-Puryear, M.D., Ph.D.
Chief
Genetics Services Branch
Maternal and Child Health Bureau
Health Resources and Services Administration

Dr. Lloyd-Puryear presented the service delivery component of the newborn screening infrastructure—namely, the new network of seven, Regional Genetics and Newborn Screening Collaboratives, and the National Coordinating Center at ACMG, Bethesda, Maryland. The HRSA grants were announced by HHS Secretary Tommy Thompson, in December 2004.

The network of Regional Genetics and Newborn Screening Collaboratives was created in response to the Children's Health Act of 2000 (Public Law 106-310). That law directs the Secretary of Health and Human Services to award grants to “enhance, improve, or expand the ability of State and local public health agencies to provide screening, counseling, or health care service to newborns and children having or at risk for heritable disorders.” In appropriations language, Congress asked HRSA to take the lead in implementing this Heritable Disorders Program.

MCHB, the Federal entity with the most responsibility for newborn screening programs, began by creating this Committee in 2003. One of the functions of the Committee (specified by legislation and in the Committee’s charter) is to provide advice and recommendations concerning grants and projects authorized under the Heritable Disorders Program.

In fiscal year 2004 appropriations, Congress specified that \$2 million from the SPRANS (“Special Projects of Regional and National Significance”) portion of the Maternal and Child Health Block Grant (Title V of the Social Security Act) should be applied to the Heritable Disorders Program. HRSA matched that amount with an additional \$2 million. MCHB is currently working with a total budget of \$4 million for the Heritable Disorders Program. Under Federal legislation, funds for this program can be used to establish a fairly encompassing list of programs related to newborn screening, but the focus is primarily on the development of the service infrastructure. MCHB is using the Regional Collaboratives as a methodology to begin implementation of the Heritable Disorders Program. It is hoped that by fostering collaborative efforts, the Regional Genetics and Newborn Screening Collaboratives will improve States’ newborn screening capacity and equalize the distribution of genetic resources within regions.

Dr. Lloyd-Puryear gave an overview of the activities proposed by each of the Regional Genetics and Newborn Screening Collaboratives. Within the first 3 years, MCHB expects each of the

seven Regional Collaboratives to achieve specified endpoints: Year 1, development of a regional coordinating plan, practice models for optimal diagnosis and follow-up and management; Year 2, development of a strategy to implement practice models; and Year 3, demonstration of public-private regional and collaborative relationships that represent a variety of health systems organizations—community health centers, health care insurers, health maintenance organizations, State-based primary care organizations, and academic institutions—for all States within the region.

The National Coordinating Center will focus on minimizing duplication of efforts, identifying best practices developed by the regions, and maximizing interregional collaboration. To date, the National Coordinating Center has proposed the following: (1) develop networks of centers of genetic services with primary care providers; (2) facilitate data information exchange and collaborate with the NIH's rare disease centers and the CDC's genomics centers; and (3) work with organizations such as the American Academy of Pediatrics and the Academy of Family Physicians to address the development of codes in the Current Procedural Terminology, and with organizations such as the Joint Commission on the Accreditation of Healthcare Organizations to bring uniformity of practice within hospitals for newborn screening programs regarding for blood samples procedures for newborn screening; and (4) encourage information-sharing projects (e.g., through telemedicine).

MCHB and the National Coordinating Center examined the proposals submitted by the seven funded Regional Genetics and Newborn Screening Collaboratives to identify several areas of shared need, as follows:

- Communication methodologies, ranging from videoconferencing to telemedicine, setting up practice models and practice relationships with genetics networks and the use of interstate satellites;
- Information management, including web-based clinical management systems suitable for telemedicine or even for satellite clinics;
- Reimbursement in general, across State lines or over a long distance (e.g., telephone consultation);
- Evaluation methodologies;
- Expansion of newborn screening, as well as the expansion of the service infrastructure needed to provide follow-up services to newborns that test positive;
- Specific regulation and legislation to allow interstate licensing for liability concerns across State borders;
- Financing, not just for the expansion of newborn screening, including the expansion of the newborn screening service infrastructure and development of new paradigms of working relationships;
- Expanding access to genetic services (e.g., via by training geneticists and primary care; increasing the diversity of trainees; and via a mechanism to more systematically address the geographic maldistribution of services).

Dr. Lloyd-Puryear noted that the National Coordinating Center for the network of Regional Genetics and Newborn Screening Collaboratives has proposed the following coordinating partners: the National Conference of State Legislators, the American Academy of Pediatrics, the Association of State and Territorial Health Officials, and the Genetic Alliance. Dr. Dougherty wondered if the National Coordinating Center was open to more suggestions for coordinating partners, such as American College of Obstetricians and Gynecologists and American Hospital Association, which Dr. Lloyd-Puryear welcomed. Dr. Howell commented that the March of Dimes Birth Defects Foundation is another obvious candidate.

The presentation raised several questions and comments. Dr. Brower asked whether carrier screening for parents and prenatal testing fall within the mandates of MCHB. Dr. Lloyd-Puryear replied, both fall within their mandates but there is no specific legislation for prenatal screening. Carrier screening would be generally considered part of the diagnostic work-up for an infant identified as screen positive in a newborn screening program and its parents.

In reply to Dr. Howell's inquiry about communication among the Regional Genetics and Newborn Screening Collaboratives, Dr. Lloyd-Puryear said, an ongoing communication process is very important and various mechanisms will be used. Also, the National Coordinating Center will bring resources to the effort. Dr. Lloyd-Puryear added, the regional grants awarded to the Regional Genetics and Newborn Screening Collaboratives are not ordinary grants. They are "cooperative agreements" that require a willingness to collaborate with MCHB and the National Coordinating Center.

Dr. Graft, suggested that the Rare Diseases Clinical Research Network join forces with the network of Regional Genetics and Newborn Screening Collaboratives. The purpose is to see how they can intersect to make activities more effective and to make knowledge of each other's activities better known to the entire community. Dr. Telfair and other committee members agreed that the relationship would provide an effort to further ensure that the results from research are translated into clinical practice.

Dr. Becker inquired about HRSA mechanisms that would enable the Committee to provide oversight of the network of Regional Genetics and Newborn Screening Collaboratives, with respect to outcomes for best practices, standardizing uniform panels of conditions across regions, and provision of services to underserved populations. Dr. Lloyd-Puryear said she presented the legislation to remind the Committee that one of its charges is to advise on the Heritable Disorders Program. The network of Regional Genetics and Newborn Screening Collaboratives does not address all aspects of the legislation, but the implementation of the Heritable Disorders Program is the first effort. MCHB also views the network of Regional Genetics and Newborn Screening Collaboratives as an opportunity to pilot standards development, and it will bring products back from the network to the Committee for review. Dr. Dougherty questioned the resources for the evaluation process. Dr. Lloyd-Puryear remarked, there is an evaluation component within each Collaborative, but with a \$4 million limit, there is no funding for overall evaluation of the Regional Genetics and Newborn Screening Collaboratives initiative. Performing such an evaluation would cost an estimated \$500,000/year. The discussion resulted in the following Committee Motion:

MOTION #1: The Committee recommends that there be an evaluation of the Regional Collaboratives Program based on a well-developed evaluation plan.

This motion was made with the understanding that there would be a plan for the evaluation and then funding would be based on the plan. As an additional step, the staff would provide the Committee with the plan for what would be evaluated that would assist the Committee in building this recommendation for what would be evaluated and how the funds would be used for an evaluation. **The motion passed unanimously.**

Newborn Screening Program Infrastructure: Evaluation and Tracking

Database for the Long-Term Follow-up of Infants Identified by MS/MS Newborn Screening

**Judith Tuerck, R.N., M.S.
Assistant Professor**

**Oregon Health and Sciences University
Child Development and Rehabilitation Center**

Ms. Tuerck gave an overview of the justification, development and effectiveness of CDC's database system for evaluating long-term health outcomes in newborns identified by MS/MS screening as having metabolic disorders. The long-term follow-up of newborns who test positive in newborn screening tests begins once the child has been diagnosed and placed on a treatment regimen—and ideally, would continue throughout the life of an individual, regardless of where in the country the individual is receiving treatment. As experience with neonatal galactosemia illustrates, some disorders and conditions thought to be benign and easily treated in early childhood may in fact have serious, long-term complications.

In 1992, the **Council of Regional Networks for Genetic Services Newborn Screening Guidelines** recognized the need for long-term follow-up of newborns diagnosed with heritable disorders and genetic diseases. It is important to do long-term follow-up to learn about: incidence/prevalence of heritable and genetic disorders; morbidity/mortality from such disorders; efficacy of treatment; natural history of disease; psychological impacts of disease; barriers to care; and the cost of care for such disorders. Long-term follow-up tracking is less than adequate, with little pooling of health outcomes information among the treatment centers around the United States. Also, any given treatment program in the United States has so few patients that the patient sample population is too small to make data analysis substantive. Now that States are implementing MS/MS screening, there is an opportunity to collect national long-term follow-up data on children identified. This, however, requires a tool for data collection, and a system to pool data to share nationally and internationally.

In 2002, CDC awarded a grant to Oregon and Iowa to develop a tool for collecting long-term follow-up data on newborns diagnosed with fatty acid oxidation disorders, organic academia disorders and urea cycle disorders. That database system is now implemented in point-of-care clinics in Oregon, Iowa, and Idaho. It is being used to collect long-term follow-up data on newborns diagnosed with 26 disorders and their siblings.

Ms. Tuerck reported the data shows that the incidence of MS/MS disorders is 1:5,300 in Iowa, 1:4,300 in Oregon, and 1:2,300 in Idaho (as compared with 1:15,000 in Hawaii). Also, the data suggest that a number of newborns in Oregon, Iowa, and Idaho were identified through the second screening test, which is routine in Oregon, as having the following metabolic disorders - carnitine transport disorders, CPT1, arginase deficiency, homocystinuria, and VLCAD (very-long-chain acyl-CoA dehydrogenase deficiency). (Other cases identified on the second test appear to be related to cutoff levels.) Ms. Tuerck remarked that it appears that not all metabolic disorders will be identified in a sample taken from a newborn in the first day or so.

The database system has the potential for other data collection applications, including the tracking of long-term follow-up of newborns with a variety of other heritable disorders and genetic diseases; evaluation of the efficacy of screening for such disorders; and evaluation of the efficacy of treatment. It may be possible to develop a National Long-Term Follow-up Center for Metabolic Disease (similar to the cystic fibrosis and the national cancer databases), to pool data from multiple centers for national analysis of either epidemiology or collaborative clinical studies to expedite the identifying of benefits and/or complications. Moreover, the database technology could be enabled to: implement automatic data collection from billing offices and laboratories; and read electronic medical records. The Northwest will consider the feasibility of implementing the database at other States in the region. The costs of collecting data, pooling and analyzing data with other centers, and system adaptability for different functions will be evaluated.

The project will investigate the use of an informed consent process to allow for more in depth data collection. A goal is to get metabolic centers to use the database system to provide data to the State or a national data pool. Additional funding would allow for the continued collection of data on these same children and any other children born.

Dr. Howell remarked he was surprised and alarmed by the positive second test results, because most States in the country do not do a second test. He asked whether there was confirmation that the first tests were negative. Ms. Tuerck replied that the cards were analyzed at outside labs using those laboratories' cutoffs and technology, and those labs verified that the tests were negative. Oregon offers a second test at approximately 2 weeks of age. Dr. Rinaldo said that Ms. Tuerck was making a very strong statement about the need for a second test. It is important to document the analytical performance in the analysis of the first test because the first test could have had an inadequate cutoff. In addition, the data should be made available for peer review.

Ms. Tuerck added that labs are struggling with cutoffs, and reported that there will be a subset of children with these diseases who will not necessarily be abnormal in the first day or so of life. This is not news for disorders, such as homocystinuria and thyroid disease. Dr. Howell concluded that the Committee should continue to discuss: (1) the need for a second screening test; and (2) what cutoff points should be used for specific tests. Discussion indicated eight States currently mandate a second screen on every baby; three States recommend a second screen and have greater than 80 percent compliance. States use data primarily on hypothyroidism and CAH, and an occasional galactosemia to point to the need for a second screen.

Newborn Screening Program Infrastructure: Integrated Information Systems

David Ross, Sc.D.

Director

Public Health Informatics Institute

Dr. Ross described the status and near-term future of integrating clinical information systems with public health information systems to improve the coordination of care for children. The Public Health Informatics Institute (PHII) believes that public health agencies and health care providers should be pushed to integrate their information systems so that providers and parents can obtain complete and accurate data sets about children in a timely way.

Dr. Ross focused on four topics: (1) the proposed National Health Information Network (NHIN) and related efforts; (2) problems in obtaining information about a child's health; (3) ongoing efforts to integrate health information systems, including pediatric electronic health records; and (4) the potential for improving health outcomes by integrating child health information systems.

This is a time of change and transformation for health care and public health. The Office of the Secretary will focus on the development of NHIN—that would allow patients, physicians, hospitals, public health agencies, and other authorized users across the nation to share clinical information electronically in real-time (under stringent security, privacy, and other protections). The hope is that the NHIN will be built over time, largely by the private sector, by linking regional clinical information systems through regional health information organizations (RHIOs) across the country. Several RHIOs and regional information systems already exist around the country, and more should be developed in the future. CDC has been working on standards for a Public Health Information Network (PHIN) that could potentially be integrated into the NHIN.

Currently, health information systems are not sufficiently integrated to allow health care providers and parents to obtain comprehensive information about a child at the point of care.

Often children found to have disorders or hearing loss during newborn screening are not followed-up and treated in a timely manner because pediatricians lack the results of newborn screening tests. Dr. Ross believes this problem can be solved as a part of the transformation of the U.S. health care information system that is beginning to take place with the move to develop the NHIN.

For the last few years, PHII has been working in collaboration with MCHB in the realm of integrating child health information systems to enable authorized users of the systems to see “at-a-glance” comprehensive information about a child’s health status and health needs. Dr. Ross believes there are four programs/systems for the newborn period that should be considered priorities for developing integrated child health information systems: (1) immunization programs; (2) newborn dried blood spot screening programs; (3) early hearing detection and intervention programs; and (4) vital records registration systems. PHII is working with a number of State public health agencies to define an integrated child health information system and to understand how to design, build, and implement such systems. This work led to the 2003 publication by MCHB and All Kids Count (AKC)/PHII, *Integration of Newborn Screening and Genetic Service Systems with Other MCH Systems—A Sourcebook for Planning and Development*—online at <http://www.phii.org/publications.html>.

PHII is working also with MCHB on the principles and core functions of integrated systems, and will release a document in the near future. It believes that no clinical or public health information system should exist without performance metrics and is developing and testing such performance measures. MCHB is providing funding through a cooperative agreement with PHII to build a community of practice composed of public health program staff and private providers to understand what integrated child health information systems can and cannot offer.

A new supplement to the *Journal of Public Health Management and Practice* summarizes recent progress toward developing integrated child health information systems, and is available from the PHII in Decatur, Georgia, or on its website at <http://www.phii.org/Files/JPHMPH%20Supplement.pdf>. Dr. Ross added a goal of the AAFP is to have at least half its members using electronic health records by 2006 and a majority of AAP members recently endorsed the adoption of electronic health records within 5 to 10 years.

Dr. Ross concluded by stating that there is a considerable amount of health information system activity currently going on. Most of it is focused on clinical systems; some of it is focused on public health systems; and a little is focused on integrating the two. Most activity is geared to programmatic rather than research functions, so it is important for researchers to be involved in the process to ensure that the health information systems are developed meet the needs of researchers. It is too early to demonstrate the impact of integrating child health information systems, but it is rational to anticipate that providing more information to users will lead to better outcomes.

Dr. Edwards asked Dr. Ross how the need for information in newborn screening could be reconciled with the need for confidentiality in an electronic information system. Dr. Ross replied that electronic technologies address systems’ security indicating who looked at electronic records, when, and potentially what they did with the information—but it will take time to become comfortable with how much less than perfect security we can tolerate. We ultimately may reach a point where people say there is more to be gained in sharing information than lost in keeping information isolated. Information technology can solve problems, but the social dialogue about this is not complete yet. Dr. Becker noted that from the States’ perspective, the Health Insurance

Portability and Accountability Act of 1996 (HIPAA) does not prohibit the reporting of public health mandated data.

Dr. Ross highlighted Rhode Island which has had a pediatric public health tracking system called KIDSNET for a decade or more. Furthermore, the State recently received a contract from the AHRQ to enter into collaboration with an RHIO in which the Rhode Island Department of Public Health participates. The KIDSNET data system will be added to the region's integrated data system, so private physicians in various hospitals and private practices will be able to gain access to data from KIDSNET. Rhode Island is an example of a State where providers will eventually be able to get a complete and up-to-date record on a child.

Committee Business - Update on the American College of Medical Genetics Report to HRSA

Peter van Dyck, M.D., M.P.H.
Associate Administrator
Maternal and Child Health Bureau
Health Resources and Services Administration (HRSA)

Dr. van Dyck reported on the status of the ACMG report to HRSA on newborn screening. The Committee reviewed a very early draft of the report at its September 2004 meeting and directed that when the ACMG report had been fully edited and completed, it should be sent to the Secretary, with a cover letter from Committee Chair, Dr. Howell, transmitting the report with the Committee's recommendation of the report. The Committee further directed that the Public Comments from the September meeting be appended to Dr. Howell's transmittal letter. The report is now final.

Dr. van Dyck said, HRSA is comfortable appending this final version of the ACMG report to the letter from Dr. Howell recommending the report to the Secretary. In comparison to the preliminary report reviewed by the Committee in September, this later version has been substantially amplified with data. In addition, the final version includes: a 200-page appendix with a two to three page description of every one of the 80 or 90 disorders discussed in the report; a discussion of the treatment; availability and cost, efficacy of treatment; benefits of early intervention and early identification; prevention of mortality; diagnosis; acute management; simplicity of therapy for each disorder; and literature cites. It also includes each disorder's score and recommendation with regard to whether a test for the disorder be contained in a core panel or not. Dr. van Dyck believes that this is the first time that this kind of full information has been available ever for even a handful of these conditions.

HRSA is now working on getting departmental permission to release the ACMG report on newborn screening for public comment. After the report's release, there will be about 60 days for a public comment period and for the Committee to comment on the report again. HHS will then take public comments and inputs from the Committee and other sources, and try to put them together into a recommendation from HHS.

Several Committee members questioned about when the final version of the ACMG report would be made available to the public. Dr. Howse emphasized that the March of Dimes would like the report in the public domain as soon as possible so people can comment on it. Dr. van Dyck explained, the report would come out for public comment as soon as they were able to gain departmental clearance, and hoped the release would occur within days/weeks. The report will be released online at the websites of the Committee and MCHB. Also, it will be mailed to organizations by request or interest. There may be a 60-day comment period, but it might be

slightly longer or shorter. An audience participant questioned whether there was a change from a 30-day public comment period to a 60-day comment period, referring to the minutes from the Committee's September 2004 meeting (p. 32):

"It was decided, given the strong interest in forwarding the report to the Secretary as soon as possible, the committee will accept and recommend the report and forward it to the Secretary immediately. There will then be a 30-day period in which electronic written forms from the public will be collected and the committee will form its comments and recommendations and forward them to the Secretary with appended public comments."

Dr. van Dyck said, the Appendix to the ACMG report sent to the Secretary with Chairman Howell's transmittal letter are the public comments from the September meeting with any additional comments received in the 30 days following the September meeting, per HRSA's interpretation of the Committee's discussion. Dr. Howell noted some confusion and differing expectations about what was to be done with the ACMG report following the September meeting. The key thing for the Committee now is to clarify its steps and move forward.

After considerable discussion, the Committee voted on and **unanimously approved** the following motions related to the disposition of the final ACMG report to HRSA on newborn screening:

MOTION #1: "The Committee directs that the ACMG report be sent immediately to the Secretary of HHS under Chairman Howell's signature on behalf of the Advisory Committee on Heritable Disorders and Genetic Diseases in Newborns and Children.

- This motion was made with the understanding that the report had already been recommended by the Committee at its September 2004 meeting. Consequently, the letter to the Secretary from Dr. Howell should say that the Committee is *recommending* the report to the Secretary.
- b. The word "immediately" was clarified to mean as soon as possible under HRSA regulations and the law.

MOTION #2: The Committee recommends that the ACMG report be released into the public domain as soon as possible for review and comment.

MOTION #3: The Committee recommends that the Committee review the ACMG report and make comments as a Committee in the same time frame as the public comments.

MOTION #4: The Committee requests an opportunity to review public comments on the ACMG report and to provide advice to the Secretary on those public comments.

January 14

Newborn Screening Program Infrastructure: Data Collection

National Newborn Screening Data Initiatives

Bradford Therrell, Ph.D.

Director
National Newborn Screening and Genetics Resource Center

Dr. Therrell gave an overview of the NNSGRC's online National Newborn Screening Information System (NNSIS)—an important component of the data and surveillance infrastructure for newborn screening. The development of the online NNSIS took about 4 years.

The NNSGRC transitioned to the online NNSIS January 1, 2005. All but one State participates in the NNSIS. Dr. Therrell showed Committee members some screens from the NNSIS Website, and described how to use the system for data entry and generating reports. It will be made available to the public through links through the NNSGRC website (<http://genes-r-us.uthscsa.edu/>).

Dr. Therrell reiterated there are several challenges with the NNSIS: (1) data reporting on newborns are neither mandated nor a defined program responsibility in most States; (2) the national data set is the result of consensus amongst newborn screening programs, and not proscribed; (3) data definitions are not consistent from one State newborn screening program to another; (4) staff shortages and turnover affect data entry into the system; (5) the quality of the data from some State programs is difficult to validate; (6) maintaining data on babies moving into a State or living on the border is often problematic; and (7) obtaining accurate data on military births is difficult.

Several Committee members applauded the development and implementation of the NNSIS by the NNSGRC, saying it would generate a tremendous amount of interest and activities for improvements in newborn screening and research. Committee members indicated they would support the Committee (or its subcommittee) involvement in the area of information management for newborn screening programs.

Dr. Becker acknowledged that the Ohio Bureau of Public Health Laboratories is using the NNSIS and believes the system will be helpful, but he doubts that States will replace their internal databases with the NNSIS. Dr. Becker outlined three challenges for the States with respect to participating in the NNSIS. States may need resources to: (1) help States with separate laboratory and follow-up services that use separate databases to communicate with the NNSIS; (2) develop mechanisms for private laboratories to report data to the NNSIS (e.g., as a pass through to the State or directly); and (3) most importantly, to ensure the quality of data reported (e.g., education and training of data entry personnel). Dr. Therrell addressed some of the challenges: (1) NNSIS has separate menus for laboratory and follow-up, so States with separate laboratory and follow-up services should have no problems; (2) NNSIS has built into the system a way for the private laboratories to download data for the State.

Dr. Rinaldo asked Dr. Therrell what the Committee might do to help with the NNSIS. Dr. Therrell emphasized Committee support for a national mandate—for States to report newborn screening data, including a mandate to report at certain times, in certain ways, and an agreement on the need for the data and the definitions. Dr. Rinaldo thinks this could be addressed by one of the subcommittees. Dr. Boyle believes that the reporting and the responsibility for follow-up reside at the state level, and that mandating reporting of data from State programs is not at the Federal level, rather NNSGRC should be a facilitator to help develop standards, centralize reporting to compare data across States, and help States with program monitoring to make State newborn screening programs work more efficiently and appropriately.

Dr. Becker agreed, but suggested it would be helpful to have a national reporting process for newborn screening, perhaps patterned on the successful model of the national infectious diseases

system, where the CDC, the Association of State and Territorial Health Organizations, and especially the Council of State and Territorial Epidemiologists produce lists of infectious diseases that all States should be reporting. Dr. Therrell clarified that he was mainly talking about expanding reporting requirements under Title V (Maternal and Child Health Block Grant).

Turning to the utilization of the NNSIS, Dr. Telfair asked Dr. Therrell whether anyone at NNSGRC has begun to analyze how the information in the NNSIS is used by States for decision making, or if not by the States, then by others who are trying to make decisions. Dr. Therrell replied that they hear anecdotally how States are using the information in the NNSIS, but suggested a need for a more systematic analysis. Dr. Therrell said the NNSGRC collates data based on what the States indicate would be useful to them and to the public; they had not considered the use of the data by other entities.

Dr. Dougherty asked whether data from the NNSIS could be used to look at the question of how many babies who get a second screening test are confirmed with secondary screening, and whether such analysis is part of the mandate of the NNSGRC. Dr. Therrell said although such analysis is not part of the center's mandate, some people have already used the screening data for analysis such as looking at second tests in the newborn screening program. Dr. Rinaldo asked about the potential for using the NNSIS data for quality improvement initiatives more generally. Dr. Therrell replied that some States have used the data in their own quality assurance efforts, but the decisions are made on a state-by-state basis. There is no mandate for the NNSIS data to be used for quality improvement initiatives. Dr. Hawkins asked how the NNSGRC collects newborn screening data on military families and families that travel overseas. Dr. Therrell is aware that a lot of the different military bases overseas have mechanisms in place to send newborn blood spot samples back to States (e.g., Colorado, Oregon, or Texas) to perform the screen. U.S. citizens who have babies abroad often have screening services provided by a local in-country program. Dr. Edwards asked whether the NNSIS is a place where doctors can access information about patients. Dr. Therrell said that although he favors such a national dataset, it's not in the near future due to privacy issues. The NNSIS obtains gross data from the States that does not link to identified patients.

Newborn Screening Program Infrastructure: Surveillance

Birth Defects Surveillance Systems

Leslie Beres-Sochka, M.S.

Program Manager

Early Identification and Monitoring Program

New Jersey Department of Health and Senior Services

Ms. Beres-Sochka gave a presentation on State birth defects and surveillance systems, highlighting the New Jersey's Special Child Health Services Registry. This registry now serves as a linchpin from the diagnosis of a birth defect to the provision of services for the child found to have a birth defect.

Currently, there are 38 operational birth defects surveillance programs in the United States; 9 additional birth defects surveillance programs are in the planning stages. Five States have no birth defects program—a situation CDC is trying to change. CDC recently surveyed the States about their ability to refer children found to have birth defects to a service system. Of the States that replied, 26 already had a system in place, 17 were planning such a system, and 3 had no plan for linking the birth defect surveillance to the service system. Among States that were linking children with birth defects to the service system, some were referring all children with birth

defects to services as New Jersey does and some referred only children with specific defects (e.g., neural tube defects or cleft palate) to services.

The Special Child Health and Early Intervention Services Program, a component of New Jersey's Department of Health and Senior Services, Ms. Beres-Sochka explained, has four organizational units:

Early Identification and Monitoring Program. The Early Identification and Monitoring Program is responsible for New Jersey's Special Child Health Services Registry, which collects data on children with birth defects and special needs. The registry contains a confidential record of all children with birth defects identified in the first year of life. The registry is allowed to use data from the State's electronic birth certificate database to amend its data and for research. The Early Identification and Monitoring Program is also responsible for New Jersey's newborn hearing screening program.

Newborn Screening and Genetic Services Program. The Newborn Screening and Genetic Services Program is responsible for screening newborns. The Newborn Biochemical Screening Follow-up Program provides information on any newborn diagnosed case into the Special Child Health Services Registry. Of the 113,404 newborns screened in New Jersey in fiscal year 2004, 6,395 were referred for follow-up, and 3,458 had abnormal results

Family-Centered Care Program. The Family-Centered Care Program is a case management program for children with special health care needs. The purpose of New Jersey's county-based case management program is to help children access family-centered, culturally competent, coordinated services for children. More than 90% of the caseload in this program is from direct referrals from the Special Child Health Services Registry. The registry tries to make a direct referral into the case management program within 10 days (or sooner if need be).

Early Intervention Services Program. In 1993, New Jersey's case managers—primary care providers or other involved professionals—began to serve as the single point of entry for the Early Intervention Services Program. The Special Child Health Services Registry gives case managers the data to referral to case management medical services, or if the children require early intervention, offer that immediately. Early intervention is voluntary and requires parental consent for participation. Any hearing loss makes a child presumptively eligible for early intervention.

New Jersey's integrated system identifies children with special needs in a timely manner; most children with birth defects are registered by the time they are 6 months old, so the link to services is made very quickly. The framework for the system is provided by separate laws and rules for the birth defects registry, the hearing screening program, and the newborn blood spot screening program that mandate not just the screening but also the reporting to the Special Child Health Services Registry. The organizational structure in New Jersey, in which four very important components of screening, surveillance and services report to the same agency (i.e., the Special Child Health and Early Intervention Services Program), facilitates communication. The integrated system is both cost-effective and efficient. Among the challenges the Special Child Health Services Registry faces are funding (currently provided by a variety of sources, including HRSA and CDC, as well as by the State and counties), ensuring confidentiality, and coping with staff turnover at birthing facilities and other health care providers.

Committee members thanked Ms. Beres-Sochka for her presentation and noted that the program in New Jersey could serve as a model for other programs. Dr. Howell commented that Ms. Beres-Sochka's figures indicated that approximately 1 in 40 newborns screened in New Jersey had an

abnormal result—a high figure. His recollection was that Mississippi data presented at the Committee’s previous meeting indicated that 1 in 300 had an abnormal result. Ms. Beres-Sochka emphasized that the data are provisional. The final data may be closer to Mississippi’s figures. Nevertheless, Dr. Howell noted, the data clearly underline the importance of the expanded programs the State has in place.

Dr. Dougherty asked whether the effectiveness of the treatment and the parent satisfaction with the case management, are being followed at all. Ms. Beres-Sochka replied New Jersey’s new electronic early intervention database system will provide a systematic way to track what treatments children are getting and whether they are improving as a result of those interventions. Dr. Howell affirmed the importance of understanding the effectiveness of treatment.

Mr. Robertson inquired about New Jersey’s definition of final case disposition. Ms. Beres-Sochka replied that it is final diagnosis. Once a newborn’s diagnosis is confirmed, the child is referred to services. Case managers keep the children in case management until the day they turn 22; after that, there is a process in place for transitioning them to adulthood.

Dr. Howse wondered if the conditions in New Jersey’s screening panel relate to the conditions recommended in the ACMG report on newborn screening. She also asked what New Jersey’s policy is with respect to screening for report-only conditions (i.e., conditions that are detectable but cannot be treated). Ms. Beres-Sochka replied that for report-only conditions, the State notifies the child’s primary care provider and parents.

Ms. Beres-Sochka clarified how the newborns that are screened get into the case management system: after the babies have been screened in the blood spot program, they get reported immediately through an electronic linkup between the lab and follow-up. The follow-up program contacts the baby’s family and physician and does not report the children to the Special Child Health Services Registry until a diagnosis is confirmed. Once the babies with confirmed disorders get reported to the Special Child Health Services Registry, they are referred to case management. When New Jersey increased the numbers of conditions in its newborn screening panel, the program proportionately increased the number of staff for the program.

Dr. Rinaldo mentioned New Jersey is one of the few States with a law that mandates metabolic postmortem screening, and asked whether New Jersey has any plans to involve the medical examiner’s office and incorporate data from that office in the Special Child Health Services Registry. Ms. Beres-Sochka said, the Special Child Health Services Registry is working with the medical examiners, to improve the process; presently the process is somewhat slower. Still, the Special Child Health Services Registry does review all death certificates of children under age 3 and requests autopsy reports from birthing facilities.

Dr. Boyle asked whether New Jersey’s Special Child Health Services Registry has data on long-term follow-up of children found to have disorders. Ms. Beres-Sochka said New Jersey does not currently have the ability to do long-term follow-up but is at a preliminary stage in developing that capacity, using money from CDC.

Committee Business: Setting Committee Priorities: Overview

Peter van Dyck, M.D., M.P.H.
Associate Administrator
Maternal and Child Health Bureau
Health Resources and Services Administration (HRSA)

Dr. van Dyck briefly explained Federal guidelines governing the formation and operation of subcommittees of federally established committees, such as this Committee:

The chair of such a Committee,—in the case of this Committee, Dr. Howell—generally has the discretion to establish whatever subcommittees he or she wants. Subcommittees are established with members of the committee. The Chair is responsible for appointing a chair of each subcommittee and may assign committee members to staff subcommittees.

Subcommittees do their work both between and during meetings. Thus, a subcommittee can get an assignment and work on the assignment by conference call, which MCHB staff will help facilitate, and then report on its work at full Committee meetings. The staff will assist with research or items that subcommittees need between Committee meetings to develop their material. If a subcommittee absolutely must meet face-to-face over a very difficult issue or for some other reason, MCHB can facilitate and help pay for that subcommittee meeting. Regular minutes of subcommittee meetings should be taken, and when subcommittees report to the full Committee, their reports are part of the official record.

At the discretion and invitation of the Committee Chair, individuals from the community or with special expertise may be invited to present to a subcommittee, contribute special expertise, or even join a subcommittee. At the discretion of the Chair and the staff, a consultant could be asked to write a paper or do some other task for a subcommittee. The activities can be paid from the Committee budget.

Dr. van Dyck also explained that subcommittees last only as long as it takes them to perform a task. Once a subcommittee's task is completed, the subcommittee can disband. Presumably, at that point, a new subcommittee would be established

Setting Committee Priorities: Discussion

Rodney Howell, M.D.

**Chair, Secretary's Advisory Committee on Heritable Disorders
and Genetic Diseases in Newborns and Children**

Resuming Committee business, Dr. Howell said that he would like Committee members to decide how many subcommittees the Committee should establish (with two to three Committee members on each subcommittee) and what those subcommittees should address. He added that he also would like to get people's preferences for specific subcommittees. Dr. Howell's impression is that subcommittees are responsible for forming their own agenda and work plan. A spokesperson for each subcommittee will present the subcommittee's plan of action at the April 2005 meeting of the full Committee before the subcommittee starts doing any work. (Dr. Howell subsequently modified this plan to expedite the process, as discussed below.)

As a framework for coming up with the specific subcommittees, Dr. Rinaldo suggested that the Committee initially use the pyramid of four "core public health services," Dr. van Dyck showed in his presentation earlier (Tab #11 in the notebook distributed to Committee members):

Direct health care services (e.g., basic health services and services for children with special health care needs;

Enabling services (e.g., transportation, translation, outreach, respite care, family support services, case management, coordination with Medicaid and Federal maternal and child health programs);

Population-based services (e.g., screening, counseling, immunization, nutrition, public education); and

Infrastructure-building services (e.g., needs assessment, evaluation, planning, policy development, coordination, quality assurance, standards development, monitoring, training, applied research, systems of care, and information systems).

Committee members made additional suggestions for subcommittee consideration: the challenges States are going to face in adopting the newborn screening panels recommended in the ACMG report; inclusion of two areas to address infrastructure: information technology and financing; longer-term issues, such as the criteria for additional selection of newborn screening tests and quality improvement; and education and training (i.e., professional training and parent education). After discussion, the Committee arrived at three subcommittees with the inclusion of several topics, along with several crosscutting issues:

LABORATORY STANDARDS AND PROCEDURES (e.g., lab standards, including case definitions/cutoffs—a high priority; quality improvement; residual blood spots);

EDUCATION AND TRAINING (e.g., education of parents and consumers, public education, health professional training); and

TREATMENT AND FOLLOW-UP (e.g., treatment protocols, evaluation of new treatments, reimbursement).

The **crosscutting issues** identified by the Committee as being of importance to all three of the proposed subcommittees were (a) evaluation of technology/tests/conditions (methodology and criteria); (b) information technology; and (c) financing.

To end the discussion, the Committee **unanimously approved** the following motion related to the establishment of subcommittees for the Advisory Committee on Heritable Disorders in Children and Newborns:

MOTION #1: The Committee shall have the following three subcommittees: (1) LABORATORY STANDARDS & PROCEDURES (which includes new technology and the addition of new tests); (2) EDUCATION & TRAINING; and (3) TREATMENT & FOLLOWUP. The crosscutting issues of evaluation, information technology, and financing should be considered by each subcommittee. Each subcommittee is to develop an agenda and work plan.

Public Comment Section

The following individuals made public statements. The written text of their statements appears in Appendix A.

Rani Singh, Ph.D., R.D. Genetic Metabolic Dietitian

Dr. Singh, noting that many children diagnosed by newborn screening are treated by immediate nutrition interventions, emphasized the urgency of training metabolic dietitians as genetics team members who can affect outcomes for children born with these disorders. She believes that well-trained dietitians can contribute in the area of research, as well as in clinical practice. Dr. Singh thanked the HRSA for funding a pilot grant program for needs assessment for continuing education needs for nutrition professionals. She also urged efforts to ensure adequate third-party

reimbursement for treatment of persons affected with metabolic disorders, particularly adults. Dr. Singh and others are in the process of establishing a formal organization for metabolic dietitians, and she feels confident that group would be willing to assist the Committee on issues of interest.

Carol Greene, M.D.

Society for Inherited Metabolic Disorders (SIMD)

Dr. Greene's statement was read by Judith Tuerck, R.N., M.S. Dr. Greene said that SIMD members appreciate the Committee's endorsement of the report "Newborn Screening: Toward a Uniform Screening Panel and System" and look forward to reviewing this report when it is available for public comment. In the meantime, SIMD urges (1) expeditious efforts to ensure that newborn screening panels across the country are rapidly expanded to identify children with treatable inborn errors of metabolism; and (2) efforts to ensure the availability of adequate resources for successful newborn screening. SIMD and its members are eager to help the Committee in any way they can.

Jill Fisch

Parent &

National Director of Education and Awareness

Save Babies Through Screening Foundation (SBTS)

Ms. Fisch made five key points in her statement. First, she thanked the Committee and other agencies working to move newborn screening in a forward and positive manner, noting the importance of collaboration among various agencies and organizations. Second, she said that although several States have recently added disorders to their newborn screening programs, there remain States that have made no move to expand their programs—she hopes that the Committee's work will help change this situation. Third, Ms. Fisch underscored the need for a national database in place to follow patients with these disorders and their response to treatment and the importance of having all States report their data to the National Newborn Screening and Genetics Resource Center. Fourth, she stressed the importance of educating health professionals about heritable and genetic disorders and increasing the number of physicians who specialize in metabolic disorders. She believes that once the health care professionals are educated, parents can become educated through their doctors. Fifth, she expressed concern about the financial situation of metabolic centers in New York and said she would appreciate any guidance that the Committee might offer on how to help the centers obtain adequate funding. Ms. Fisch closed by thanking the Committee for its support and efforts.

Jana Monaco

Parent &

Board Member

Organic Acidemia Association

Ms. Monaco—who has a son with severe brain damage due to late diagnosis of isovaleric academia and a daughter who is healthy because of early detection—noted that States increasingly are aware of the fact that they need to make changes, but babies with metabolic disorders are still dying or suffering because their disorders are not detected and treated early. Recommending a universal panel of newborn screening tests, therefore, is just the first step in a long process to ensure effective testing and treatment of babies with metabolic disorders. Ms. Monaco urged the Committee to involve parents and organizations like hers in efforts to help raise awareness, support research, educate professionals, and implement long-term follow-up of babies found to have metabolic disorders. She closed by thanking the Committee on behalf of the Organic Acidemia Association for moving forward to develop a universal newborn screening program that will reduce disparity among States and ensure quality and effective testing and treatment for babies and children in the United States.

William Slimak
Vice President of Operations
Pediatrix Screening
Pediatrix Medical Group

Mr. Slimak noted that he would be making a statement for Pediatrix in place of Philip Vaughn, M.D., who had laryngitis. Mr. Slimak stated that Pediatrix Medical Group is a leader in hearing screening and metabolic screening and was instrumental in developing tandem mass spectrometry. Pediatrix believes that the American College of Medical Genetics report on newborn screening is important to adoption of broader scope of screening services and is pleased that it is about to be made available to the Secretary of Health and Human Services and the public. Pediatrix has operational, clinical, and technical expertise related to newborn screening and hopes that it can contribute to the work of the Advisory Committee on Heritable Disorders and Genetic Diseases in Children and Newborns and its subcommittees.

Micki Gartzke
Parent &
National Director of Education and Awareness
Hunter's Hope Foundation

Ms. Gartzke thanked the Committee and its partners for their hard work and commitment. She noted that the Hunter's Hope Foundation, which started in 1997 with a mission to increase public awareness of Krabbe disease and other leukodystrophies, is now committed to working to ensure that all babies born in the United States receive newborn screening for all the diseases for which tests are available. Ms. Gartzke and her colleagues anticipate that the public release of the American College of Medical Genetics report will be a major step forward. Ms. Gartzke said the priorities of the Committee and its subcommittees should be driven by the interests of what is in the best interests of the children. Thus, a key priority should be to educate expectant parents and physicians and medical students about newborn screening and disorders. Other issues of importance include improving the speed and quality of newborn screening, providing effective treatment and follow-up care to children with disorders, expanding data collection to find out what is working best so it can be applied nationally, expanding funding for newborn screening efforts, etc. Ms. Gartzke ended her statement by saying that the Hunters' Hope Foundation will very gladly do whatever it can to help the Committee do its job.

Joyce Hooker
Executive Director
Mountain States Genetics Network

Ms. Hooker noted that the Mountain States Genetics Network is one of the new Regional Genetics and Newborn Screening Collaboratives being funded by HRSA—specifically, the Region 6 Genetics Collaborative Center (which covers Arizona, Colorado, Montana, New Mexico, Texas, Utah, and Wyoming). One of the most important and productive activities of regional networks, Ms. Hooker said, is providing the opportunity to network, because networking—whether formal or informal—is the first step to collaboration and the coordination of services. The Mountain States Genetics Network is committed to providing opportunities for face-to-face networking. Ms. Hooker ended by saying she hopes that the Committee will regard the Regional Genetics and Newborn Screening Collaboratives as a knowledgeable and fervent resource.

Peter Sybinsky, Ph.D.
Chief Executive Officer
Association of Maternal and Child Health Programs (AMCHP)

Dr. Sybinsky began his statement by explaining that AMCHP represents State and territorial public health leaders who administer statewide programs to improve maternal and child health. Testing newborns for metabolic disorders, Dr. Sybinsky said, is essential to giving every child a strong start in life. As the Committee moves forward with its work, AMCHP urges it to recognize

that effective newborn screening is more than just testing—it is an entire system that includes parent and doctor education, medical referrals and follow-up, and ongoing support for a child’s medical care. AMCHP also urges the Committee to recommend the provision of adequate Federal resources to support State newborn screening systems. Although States rely significantly on the fees charged to families or hospitals to finance testing and laboratory costs, the Maternal and Child Health Block Grant usually pays for follow-up services and specialty care when it is not available through Medicaid, the State Children’s Health Insurance Program, private insurance, or other sources of funding. For that reason, AMCHP calls on the Committee to recommend adequate funding for these programs. Dr. Sybinsky concluded by saying that AMCHP thanks the Committee for its efforts and looks forward to providing any assistance to the Committee that it can.

Committee Business: Setting Committee Priorities, continued

Rodney Howell, M.D.
Chair, ACHDGDNC

Dr. Howell made the following assignments to subcommittees, with individual preferences taken into account.

SUBCOMMITTEE ASSIGNMENTS
LABORATORY STANDARDS & PROCEDURES Chair: Dr. Brower Members: Dr. Alexander, Dr. Coggins, Dr. Rinaldo and Dr. Howell
EDUCATION & TRAINING Chair: Dr. Howse Members: Dr. Edwards, Dr. Becker, Dr. Hawkins, and Dr. Collins
FOLLOWUP & TREATMENT Chair: Dr. Boyle Members: Dr. van Dyck, Dr. Dougherty, Mr. Robertson, and Dr. Telfair

Committee discussion noted that if the subcommittees worked on their agendas at the April 2005 meeting, there would be no subcommittee work in the interim period. Dr. van Dyck laid out a proposal to have subcommittee agendas reviewed and approved electronically in the interim. As Chair, Dr. Howell could specify that the subcommittees are to form an agenda within 30 days, for example. Upon receiving the subcommittees’ proposed agendas, the staff at MCHB could e-mail them out to the full Committee, and there could be a comment period of a week or some other period. Once the Chair approved a subcommittee’s agenda, he would notify the subcommittee of his approval to proceed with its work. Dr. Howell agreed that communicating by phone and by e-mail to expedite the review of the subcommittees’ agendas was a good idea and told Committee members to submit their agendas or actions plans by the end of February.

The establishment of a Committee listserv was discussed and it was decided to continue to use the current Committee listserv for communication. In the interim, any agendas or action plans that Committee members would like distributed to other Committee members should be sent to Dr. Lloyd-Puryear. Dr. Howell agreed that Dr. Lloyd-Puryear would circulate subcommittee agendas and action plans to the full Committee and keep records on them.

There was consensus that the new subcommittees should submit a proposed agenda and a priority action plan to Dr. Lloyd-Puryear for distribution to the full Committee by end of February 2005. Dr. Howell would approve these by early March. As noted by Dr. van Dyck, the priorities of the subcommittees may drive the agenda for the Committee's next meeting on April 21-22, 2005.

Dr. Howell concluded the discussion by asking Committee members to suggest issues of interest for the April 21st meeting—regardless of what comes out of the subcommittee groups. Committee members made the following suggestions:

- *Crosscutting issues.* The Committee should flush out the crosscutting issues that are to be addressed by the subcommittees—i.e., evaluation, information technology, and financing. Outside experts could be invited to talk about these issues. Dr. Howell said that Dr. Lloyd-Puryear could start working on inviting experts/speakers. Committee members should let Dr. Howell know if they have people they want to recommend.
- *American College of Medical Genetics (ACMG) report on newborn screening.* An agenda item for the Committee's next meeting and subsequent meetings is a continuing status report on the ACMG report on newborn screening produced for HRSA. By April, the report will have been released to the public. Dr. Howell agreed that having a report update as an agenda item would be appropriate. Dr. van Dyck said that MCHB staff would be happy to report on the status of the report at each meeting. Discussion indicated that ACMG Executive Director, Michael Watson, Ph.D., be invited to present on those aspects of the report that the Committee hasn't already considered, along with the report's recommendations. Dr. Boyle reminded Committee members that they had voted on several motions pertaining to the report on the previous day.
- *Legal issues related to newborn screening.* Dr. Rinaldo and Dr. Edwards suggested that perhaps the Committee should pay some attention to the legal issues related to newborn screening programs. Dr. Howell thought the Committee could benefit from having consultants speak on legal issues relevant to newborn screening. Given the many items already on the suggested agenda for the April meeting, Dr. Hawkins suggested that the legal issues be deferred for a subsequent meeting; in the interim, the three new subcommittees could start making a note of important legal issues and give them to Dr. Howell. Dr. Howell agreed that this approach would be helpful.
- *Research.* Dr. Rinaldo suggested research as an agenda item for later. Dr. Howell said that as the subcommittees work toward some of the immediate solutions, areas where the knowledge is incomplete will be evident. That will obviously suggest a great research agenda.

Dr. Howell reminded members of the public that it is important to let the Committee know in advance that they are coming to make a public statement. It is also helpful if they submit a written statement to ensure that their statement is accurate in the record.

The next three Committee meetings, Dr. Howell noted, are scheduled for the following dates, and all will be held in the Rotunda Ballroom, 8th floor of the Ronald Reagan Building and International Trade Center, Washington, DC:

- April 21-22, 2005
- July 21-22, 2005
- October 20-21, 2005

Dr. Howell said that Committee members should consider that the aforementioned dates are firm. Dr. Howell thanked the members, speakers, and audience members for participating, and concluded the meeting.

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We certify that, to the best of our knowledge, the foregoing meeting minutes of the Advisory Committee on Heritable Disorders and Genetic Diseases in Newborns and Children are accurate and correct.

/s/ _____

R. Rodney Howell, M.D., Ph.D.
ACHDGDNC, Chair

/s/ _____

Michele A. Lloyd-Puryear, M.D., Ph.D.
ACHDGDNC, Executive Secretary

These minutes will be formally considered by the Committee at its next meeting, and any corrections or notations will be incorporated in the minutes of that meeting.

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Professor

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APPENDIX A: WRITTEN PUBLIC COMMENTS

1. Rani Singh, Ph.D., R.D.—Genetic Metabolic Dietitian
2. Carol Greene, M.D.—Society for Inherited Metabolic Diseases [statement read by Judith Tuerck, R.N., M.S.]
3. Jill Fisch—Parent & National Director of Education and Awareness, Save the Babies Through Screening Foundation
4. Jana Monaco—Parent & Board Member of the Organic Acidemia Foundation
5. William Slimak, Vice President of Operations, Pediatrix Screening, Pediatrix Medical Group
6. Micki Gartzke—Parent & Director of Education and Awareness, Hunter's Hope Foundation
7. Joyce Hooker—Executive Director, Mountain States Genetics Network
8. Peter Sybinsky, Ph.D.—Chief Executive Officer, Association of Maternal and Child Health Programs

Rani Singh, Ph.D., R.D.
Genetic Metabolic Dietitian
Statement to the Advisory Committee
on Heritable Disorders and Genetic Diseases in Newborns and Children
January 14, 2005

Mr. Chairman:

Thank you for this opportunity to speak as a metabolic dietitian. I want to open my comments by acknowledging that a majority of the diseases diagnosed by Newborn Screening (NBS) recommended by the American College of Medical Genetics (ACMG) panel are treated by immediate nutrition interventions. Therefore, the qualifications and the role of a metabolic dietitian as a genetics team member can have a great impact on the outcome of these children. I wanted to emphasize the urgency for the need to train and support dietitians in the field. (I was very excited to note Nutritionist clearly listed under Direct Services in Peter Van Dyck's presentation.)

This becomes even more critical in the face of shortage of biochemical geneticists while expanding newborn screening. I have created an International List-Serv for metabolic dietitians and now we have close to 200 members. Participating members continuously express confusion with treatment protocols and lack of understanding with the standards of care which lack evidence-based research and treatment protocols. I feel well trained dietitians cannot only contribute in clinical practice but also in the area of research.

I also want to thank HRSA for funding a pilot grant in Region 3 for needs assessment for CE (continuing education) needs and development of the first educational module for nutrition professionals. I do want to take this opportunity to urge that efforts to assure availability of qualified dietitians in the metabolic centers and the adequacy of nutrition resources like medical foods and low-protein foods are available for treatment. Lack of third party reimbursement continues to threaten the availability of treatment in some States for affected persons particularly those who have now entered adulthood.

We are in the process of establishing a formal organization for metabolic dietitians to meet our needs and feel confident that group would be willing to assist the committee with any efforts focused in this direction.

Carol Greene, M.D.
Society for Inherited Metabolic Disorders
Statement to the Advisory Committee on Heritable Disorders and Genetic Diseases in
Newborns and Children
January 13-14, 2005

Thank you, Mr. Chairman, for the opportunity to speak, and thank you to the Committee for your efforts on behalf of those with inborn errors of metabolism and other genetic disorders.

The Society for Inherited Metabolic Diseases (SIMD) is dedicated to improving scientific and public understanding about inborn errors of metabolism, and to promoting advances in the identification and care of those affected by inborn errors of metabolism. Members of the SIMD are scientists, physicians, nutritionists, nurses and other health professionals working in patient care and in research, in the laboratory and in the clinic, in academia, in public health, in private medical systems and in the biotechnology industry.

SIMD members provide diagnostic and treatment services to individuals of all ages with inherited metabolic disease to minimize the risks of disability and death. SIMD members play a prominent role in the diagnostic follow-up and treatment of children detected by newborn screening with inborn errors of metabolism. From this perspective, we very much appreciate your endorsement of the report “Newborn Screening: Toward a Uniform Screening Panel and System” at your last meeting, and we support your efforts to expand newborn screening. We understand from presentations at your meeting that this endorsement is an important step along a path to expanding the panel of conditions for which newborns in the United States are screened, and to do so with uniformity. We look forward to reviewing this report when it is available for public comment. In the meantime, we wish to take this opportunity to urge:

- Expeditious efforts to assure that newborn screening panels across the country are rapidly expanded to identify children with treatable inborn errors of metabolism.
- Efforts to assure availability of adequate resources for successful newborn screening and follow-up. The diagnosis of a biochemical genetic disease in an infant detected through newborn screening should be confirmed in a qualified diagnostic laboratory, and immediate and long-term treatment should be available from qualified and experienced experts in inborn errors of metabolism. Mechanisms need to be in place to adequately fund all aspects of newborn screening, and to fund the treatment of inborn errors of metabolism in those who are identified by newborn screening. Funding needs to be assured for education, testing, reporting of results, confirmation of abnormal screening results, diagnosis, comprehensive long-term treatment and evaluation of patients, and outcomes evaluation of newborn screening programs and practices.

Again we thank you for this opportunity to speak, and want to assure the Committee that the SIMD and its members are eager to help you in your efforts on behalf of the people we both serve.

Jill Levy Fisch
National Director of Education and Awareness
Save Babies Through Screening Foundation
Public Comments
January 14, 2005

Good afternoon. It is a pleasure to be here today and an honor to have the opportunity to address the committee.

My name is Jill Fisch. I am the National Director of Education and Awareness for the Save Babies Through Screening Foundation. I would like to acknowledge and say thank you to the committee and other agencies that are hard at work to move newborn screening in a forward and positive manner.

Collaboration between various agencies and organizations is essential. Newborn screening is not just an issue of the actual screening and gives rise to other issues which need to be addressed. Some of the issues are education, financing, data collection, evaluations and infrastructure. Other issues of great importance are linking identified children to services, ancillary services, manpower, money for treatments and parental difficulties.

Some states are finally paying attention to newborn screening. Since the last meeting, Alabama, Connecticut, Louisiana, Michigan, Minnesota, New York, South Carolina, North Carolina and Georgia have added disorders to their programs. New York will be screening for all disorders by the spring and Florida is going to begin adding disorders in February for a total of 25 added by the end of 2005. It is truly wonderful to see such progress being made. However, there are states where there is no move to expand and it is my hope that upon Secretary Thompson's acceptance of this committee's recommendations that things will change. For those viewing this issue from a financial perspective versus quality of life Colorado's newborn screening states that "identifying just two cases of PKU and 12 cases of hypothyroidism this year will save \$2.5 million in lifetime costs for institutional care and special education." Delaware's states that "newborn screening, together with rapid diagnosis and treatment prevents mental retardation, illness, and death in newborns. It also saves millions of dollars in treatment, home and institutional cost. For every \$1 spent on newborn screening, \$9 in medical care and treatment costs are saved—resulting in a national savings of \$36 million every year." Since the last meeting approximately 530 unscreened babies with disorders have been born. I now view these babies as ticking time bombs. Time is of the essence.

There has been much discussion regarding the efficacy of treatment of certain disorders. One of these debated disorders, SCADD, has greatly affected my family. I have seen first hand that treatment can be effective. My father has been receiving treatment for several months and is finally able to manage the stairs in his home without difficulty. We need to have a national database in place to track patients with these disorders and their response to treatment. This is the only way we will be able to see what treatments are effective. All states should be reporting their data to the National Newborn Screening and Genetics Resource Center, so that needs are easily identified and support can be given. I am not quite sure I understand why NY is the only state in the country that refuses to produce their data. This concerns me, greatly. I am hopeful someone on the committee can help me understand why this is occurring.

I think education needs to be the number one priority as the subcommittees are developed and prioritized. We need to educate health care professionals about these disorders. The number of knowledgeable sub-specialists is inadequate. My younger son had to wait 6 months to see a neurologist who specializes in metabolic disorders. Raising awareness in the health care field will allow these professionals to choose sub-specialties they had not been aware of before.

We also need to educate the health care professionals about the screening itself and the impact it has on families. One way to educate health care professionals about screening and the impact of these disorders is for Professors to provide the education to their students. Families can be a great resource in the teaching process by coming to speak in classes. Hearing families speak would have a great impact and would ensure that students actually remember the lessons. Almost all Professors should be able to arrange for families to come and speak. Many of these Professors are doctors who also treat patients, especially those affiliated with children's hospitals in major cities. Duplicate this effort nationwide and you have education going on. The other avenue for education regarding screening and disorders is free online CME. The Save Babies gets 20,000 visits per month from professionals and parents all over the world. We have asked Pediatrix Screening to develop online CME in light of all the developments in newborn screening and have assured them that we would be happy to put it on our or provide a link to CME on their site. This is now in process. We would be open to working with any other agency or organization interested in pursuing this avenue of education also.

Once the appropriate people work together and provide education to health care professionals, parents can become educated through their doctors. This is happening already in California. California is a shining example of effective parental notification and should be followed by others. The California Department of Health has circulated and continues to circulate the Save Babies pamphlet to the offices of every appropriate doctor and midwives with an insert specific to their state newborn screening program. We are receiving a steady stream of phone calls from the offices and pamphlets are being ordered in record numbers. They are then distributed to pregnant women. We also have been getting inquiries from parents themselves who have been directed to us by their health care professional.

Lastly, I need to raise an issue greatly impacting the metabolic centers in NY. I am thrilled about the expansion of the newborn screening program in NY. However, the program is being implemented without a fee. As a member of the Advisory Board of the metabolic center in the lower Hudson valley, I have been asked to help address this issue. The metabolic centers in NY are unable to properly and adequately meet the need of their patients without the fees generated from screening. There are no funds for repeat testing, follow-up, formula and other needs of the families. I have reached out to the Governor's office and the Department of Health to no avail. Our local center is now attempting to raise money through private donors to meet the needs of the center. This is unfortunate as the doctors at the centers should be able to just focus on their patients and not have deal with the aspect of raising funds. I am working with Legislators in an attempt to rectify this situation and obtain the much needed funds for the centers. Any guidance that could be given to me by the committee regarding this matter would be greatly appreciated.

Thank you again for all of your hard work. I look forward to seeing these issues move forward at the direction of this committee and offer my assistance to the committee in any way it sees fit. We all have the same goal. We are all in this together. Thank you once again for the opportunity to speak today.

Jill Levy Fisch
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Jana Monaco
Parent & Board Member of the Organic Acidemia Foundation
Public Comments
January 14, 2005

Good afternoon and thank you again for the opportunity to come and speak on behalf of Expanded Newborn Screening. I am the parent of two children with Isovaleric Acidemia, Stephen with severe brain damage due to late diagnosis and Caroline who is a healthy normal two year old thanks to early detection. I wish to thank each of you for the dedication that you have committed to NBS. We are all aware of the fact that it has not been an easy process, but you have diligently worked to finally get a report completed and prepared to send to the Secretary. The mere fact that a report is coming has helped move things along in the state of Virginia as well as other states. As pointed out yesterday, states are aware of the fact that they need to make changes and Virginia is moving in that direction. With all due respect, as the parent of a child severely disabled due to lack of Comprehensive NBS, I have to disagree with Mr. Ross' statement from yesterday, stating that our NBS program is a highpoint in the American medical system. If it was, we wouldn't be here today. It's good but far from stellar. It no doubt teaches us that we have a long way to go. A recent example of this important fact is a baby names Joseph in Norfolk, whose grandmother has connected with me to share his story and seek advice. He is a 4-month-old baby diagnosed 3 ½ weeks ago with MMA. His diagnosis came after weeks of visits to the pediatrician with concerns only to be brushed off and attributed to the concerns of a young, single mother. Their lack of health insurance also played a role in the lack of urgency with his care. Like Stephen, Joseph has been on a ventilator and the family has been told to consider turning off life support.

Over the past few months, I have spoken to the Virginia Genetics Advisory Committee, and a local town hall meeting which has helped Virginia adopt bills in the House and Senate to expand Newborn Screening, to be consistent with the panel recommended by the American College of Medical Genetics, in the soon to be released report. The regulations must include follow-up and referral protocols and necessary provisions to implement the NBS program and any services available to the infants through the Children with Special Health Care Needs Program. To help support the bill, I have been asked to speak and share my children's story at the General Assembly sessions when the bills are read. I also had the privilege of speaking at the Northern Virginia Pediatric Society lecture in November. This was a great opportunity to educate physicians who are not up to speed with these disorders. In all of my efforts, I have learned that people do want to know about this subject and help bring about the necessary changes to provide our country's children with a healthy start in life. It just isn't acceptable to continue to hear medical personnel admit that they know little about inborn errors of metabolism. The emphasis should be on education in this process of expanding NBS. It should be the number one priority in your subcommittee development. Each time one of our children is hospitalized, we parents educate one more medical staff person on these disorders. Recommending this universal panel is just the first step in a long process. Like me, other parents around the country are committed in their own states to help this process.

Although I am not someone of your professional expertise, I do have more hands on experience with living with these disorders like the rest of us parents. This brings with it, a parent perspective on the subject that is vital in the development of this Newborn Screening program. Various models already exist within the health sector. The concept of some sort of data base or communication system for health care professionals is a critical component as well. There is such a concern regarding privacy issues, but I can assure you that we lost our privacy the day Stephen went into crisis. When Stephen was first diagnosed, we had requested information about

Isovaleric Acidemia. The physician tried to provide us with what she could to include some cases of IVA to read. They were old cases off the internet and did not provide us with much in the way of understanding the prospects of a life with IVA. It wasn't until we were connected with the Organic Acidemia Association that we learned a great deal about IVA. That is where we discovered that Stephen was not the only one diagnosed beyond the newborn period and that there were older IVA children and adults living with IVA. This is where follow-up information can be very useful in education. We were introduced to other families who had a great deal of information to share with us in regards to raising a child with an inborn error of metabolism... information that we did not receive from the physicians. As you form your subcommittees and seek to find ways to do long term follow-up and possibly develop informational data bases, use us parents in some sort of capacity. Our organizations are full of parents willing to help raise awareness, support research, and provide assistance with educating others. Many come with their own level of professional expertise that compliments our organizations. We are a place to begin this aspect of the NBS process as well as other components of the process. We are people that take initiative in case you haven't already noticed, and have already relinquished a level of privacy for the sake of NBS and to connect with others. My husband and I along with three other IVA families have developed a called IVASupport.org whose goal is to be an outlet and resource solely for families of already diagnosed and newly diagnosed IVA children. It contains stories and photos of children with IVA, a detailed definition of IVA, NBS information articles and studies on IVA by physicians, and a physician and dietitian on board for questions. We have provided a letter to our metabolic clinics and pediatricians to share with any new IVA families. As a parent, this is something that I wish had been available to us upon Stephen's diagnosis. This is just an example of how resourceful parents can be. Again, I stress that you utilize us. We are a great resource for understanding the needs of raising children with inborn errors of metabolism and have much to contribute.

In closing, I speak for the Organic Acidemia Association in thanking you for moving forward to develop a Universal Newborn Screening Program that will reduce disparity among states and ensure quality and effective testing and treatment for babies and children in the United States.

William S. Slimak
Vice President of Operations
Pediatrix Screening
Public Comments
January 14, 2004

Dr. Puryear, Secretary to the Advisory Committee on Heritable Disorders and Genetic Diseases in Newborns and Children (ACHDGDNC)

Following public comment submitted on January 14th to the ACHDGDNC meeting by Bill Slimak, Vice President of Operations, Pediatrix Screening

Members of the Committee and other attendees, thank you for the opportunity to address you today. My name is Bill Slimak, Vice President of Operations at Pediatrix Screening.

Qualifications – extensive background in healthcare operations....Clinical Chemist by training – over 30 years of Pharmaceutical Operations – licensed Biologics.

Proud to be a part of this enterprise where we can do so much good for future generations. Pediatrix Medical Group's mission is to improve the lives of newborns. Screening is an important part of that mission. Pediatrix Medical Group has been a leader in Newborn Hearing Screening and now Newborn Metabolic Screening.

Pediatrix Screening was instrumental in developing the application of Mass Spectrometry to Newborn Screening and now has 10 years and over 2 million babies experience....operationally, technically and clinically.

Pleased that the American College of Medical Genetics report will now be made available to Secretary and to public. Eagerly anticipate feedback from the relevant departments and public at large to help inform future direction. This will be an important step in encouraging the adoption of a broader scope of screening services, through whatever means best meets the needs of individual programs.

Pediatrix Screening and Pediatrix Medical Group anticipate that through our participation in upcoming subcommittees we will be able to substantively contribute given our experience, and the depth and breadth of expertise:

- Screening technologies – unparalleled experience with the combination of technologies we use today, primary and second tier;
- Research – Research & Education continuing to develop future directions for screening to improve identification of affected newborns;
- Data management – vast experience with developing and managing multifaceted patient data, 250,000 deliveries/year;
- Clinical trials – Genetic Counselors to provide specialty patient communication, data warehousing, and clinical trials management experience ideal for development of future of newborn screening, where we have a much greater understanding for the diseases we identify and treat through newborn screening systems;

- Standards – We also recognize the importance of standardization where possible. Pediatrix Screening actively participates in important standardization committees to ensure patient safety, and build continued confidence and capability into the advanced technologies used in newborn screening;
- Operational control – We have extensive experience in logistics management, process control and continuous improvement to ensure Quality and Efficiency.

In closing, we have the operational, technical and clinical expertise to help this committee in reaching our mutual goals ensuring that newborn screening is available to all.

These issues are complex and states, parents and primary care physicians will be looking to all of us in the newborn screening community to help. To omit Pediatrix Screening from this process does not further the goals or mandates of this committee. For these reasons, we respectfully request the opportunity to participate as a member of the proposed subcommittees.

Thank you again for the opportunity to speak on this important issue. Pediatrix Screening looks forward to an increasing our participation in this process.

Micki Gartzke
Director of Education and Awareness
Hunter's Hope Foundation
Public Comments to HHS Advisory Committee on Heritable Disorders and Genetic
Diseases in Newborns and Children, Ronald Reagan Center, Washington DC
January 14, 2005

Good Afternoon Mr. Chairman and Members of the Committee. Thank you and the many collaborating agencies and partners for your work on behalf of children and families in the U.S. This joint effort is invaluable to the future of American families. It is my pleasure and an honor to be able to share my comments with you today!

My name is Micki Gartzke, I am the Director of Education and Awareness for the Hunter's Hope Foundation; we are committed to all children having equal access to all available newborn screening. Along with the many thousands of families we work with, who have gained awareness of the need to improve NBS on a national basis, we applaud you for your hard work and commitment towards fixing this huge national public health problem.

We are excited about the progress that will come with the forward movement of the American College of Medical Genetics report. With more and more involvement and greater partnership on the variety of issues still to be solved, more children's lives will be saved, and that is a great asset for America!

We think the priorities for the sub-committees should be driven, first and foremost, by what is in the best interests of the children. And that, right now, is education. Further resolution of the agenda items will bring better and faster newborn screening and consequent follow-up care to children, and that is what is at stake, providing effective treatment to those children identified through newborn screening. With we expand data collection nationally, we will find out what's working best so we can apply it broadly across all the states to help health care professionals do their jobs better. With expanded funding and funding strategies, equitability will become a reality and we will realize more effective treatments. With ALL states reporting their data to the collective resource of the National Newborn Screening and Genetics Resource Center, those states with needs will be identified. Support can flow where it is needed. With expanded infrastructure, opportunities will be more broadly available linking the children to the necessary services that are of the utmost importance in this process. I continue to hear of children having 6 month waits to see pediatric neurologists. I continue to hear of medical centers not having adequate resources to give the proper care. These issues need to be fixed so the children can receive the best care possible. We need to get real NBS educational information into the hands of expectant parents so they can ensure that their newborn is receiving the best possible option for newborn screening! We need to engage the obstetric providers to provide this information. We need to educate our medical students about newborn screening. With more education available at the medical school level, the importance of the entire newborn screening process will generate more physicians filling the service gaps that exist today. This education can be strongly supported by partnership, including parents who can provide real life experiences. I know that this type of education makes a difference as I have spoken to many first year medical school classes on the issue of the pediatric hospice because of my experience of having a young daughter needing that valuable type of care.

We have seen that the states are listening and some have made progress. Thank you Dr. Therrell for your wonderful presentation and thank you Jill Fisch for highlighting some of this information in your comments. Just this morning I read new NBS bills in Kentucky, this is definitely another good sign of progress. Still, many states that lag. Not only in the area of providing adequate newborn screening, but even in getting the word out in an effective and meaningful way to parents. For many parents this helpful information is dismally unavailable. We applaud California for making a real effort to get vital information into the hands of all relevant physicians and midwives. These healthcare professionals can then get the word to parents in a timely fashion to help them make the choice of screening supplementally while CA works to get up to a higher standard of care in newborn screening.

It seems to me and other advocates that there are still many questions that need to be answered and I know you all will work hard to find the answers to improve newborn screening standards nationwide. A couple of questions that come to “top of mind.”

Where are the parents still having difficulties? Are we spending enough money on treatment? Are there adequate sub-specialists geographically located to provide the necessary services? What can be done to motivate the still resistant states to expand their services? We will do whatever we can to help and we are looking for your guidance on how we may expand our advocacy efforts. We look forward to your advice.

Hunter’s Hope started as a Foundation in 1997 with a Mission to increase public awareness of Krabbe disease and other leukodystrophies to increase the likelihood of early detection and treatment of these diseases. Of course, we know that access to the life-saving treatment lies in the hands of expanded and universal newborn screening. We quickly realized that it was imperative to advocate on both state and Federal levels across the country to ensure that all babies born in the U.S. receive newborn screening for all diseases for which there are tests available. Originally, we started out to save the lives of children born with Krabbe and related Leukodystrophies. And now, through our strategic planning, our fight for early identification has expanded to all diseases that have newborn screening tests, to help save these children’s lives in America.

To date, in addition to providing nearly 4 million dollars toward Leukodystrophy research, Hunter’s Hope has developed a universal newborn screening campaign. This campaign includes multiple meetings with Federal legislators discussing newborn screening, we have asked for additional funding to support newborn screening. We have been active in many different states, writing to health officials and legislators, supplying volumes of information to our parent-based newborn screening taskforce, supporting their efforts on the ground to get their individual states to improve their newborn screening program. Jim Kelly, one of our founders, has made many speeches around the country on newborn screening, including his keynote speech at the World Congress on Disabilities this past October in Orlando, FL, to cite one specific effort. We have developed a “pilot” brochure highlighting the need for expanded newborn screening. It has been mailed it to thousands through a qualified mailing list. These types of efforts continue to expand on a daily basis. We continue to write articles about the need for expanded newborn screening in each of our Hunter’s Hope Foundation newsletters, with each mailing going to approximately 40, 000 households. We reach out to the media to publish more awareness information on newborn screening. We have met with state officials in New York, alongside Jill Fisch, working to get New York to expand is program. We are in the process of supporting a pilot program in New York State for newborn screening for 5 Lysosomal Storage disorders. We are advocating gaining additional states in this pilot program. Wisconsin has indicated a preliminary interest in participating. Next month I will be in North Carolina seeking its participation. We are providing bloodspots of affected Krabbe children and healthy siblings for this pilot program. We are

reaching out to other lay advocacy organizations to expand this support. We plan to meet with various transplant doctors from around the country, as we host at our 8th annual Medical and Family Symposium in Western New York this July to help expand treatment for newborns of Leukodystrophies and Lysosomal Storage Disorders.

We know the children can be saved. We know that you are committed to make that happen as quickly and effectively as possible. We will do whatever we can to help you do your job. We hope you will ask us to help. Thank you!

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**Testimony Given Before the Secretary's Committee on Heritable Disorders and Genetic Diseases in Newborns and Children
January 14, 2005
Washington, D.C.**

First I would like to thank the Genetics Branch and Hereditary Diseases Program for funding the regional collaborative centers. I have been the Regional Coordinator for the Mountain States Genetics Network since 1985. Over the years the regional genetics networks produced numerous pamphlets, brochures, booklets, professional journals and articles, collected data on newborn screening and clinical services provided, conducted surveys, prepared reports and position papers, created legislation, and set up clearinghouses for materials just to name a few items. But the most important and productive activity has always been the opportunity to network. This is the backbone of the Mountain States Genetics Network now the region 6 genetics collaborative center.

Networking can take on many patterns and designs and is the first step to coordination and collaboration. For instance, there is what we refer to as formal and informal networking: formal being a designated time and place and informal anytime, anywhere. It can take place on the phone, Internet, or via the many telecommunication systems available. But the most effective and valuable means of networking is the age-old face-to-face meeting. Each year the MSGN asks its members and Annual Meeting attendees which activity or facet of the MSGN is the most valuable to them and it is always the mid year committee meetings and Annual Meeting. The MSGN has seven multi-disciplinary committees each on a particular issue. All committees, whether it is the Consumer Issues Committee or the Laboratory Practice Committee, is comprised of consumers, physicians, genetic counselors, laboratory staff etc. The MSGN is committed to providing each committee a full day face-to-face meeting in the winter and a three-hour segment at the Annual Meeting in July. This affords the committee members time and opportunity to meet and know their peers and others in the region, to sort out and discuss issues and problems, to plan strategies and activities the committee will carry out and share data and ideas in a comfortable, non-confrontational environment. This doesn't just happen on its own. It takes commitment and work from those attending and the staff. But the benefits are well worth the resources it takes and we get a big bang for our bucks. It is important in this great age of electronic technology and communication that we not forget the human element. After all, that is what we are all about. Again thank you for your generous support and we hope you will look to the regional collaborative centers as a knowledgeable and fervent resource.

Respectively submitted by,
Joyce Hooker, Executive Director
Mountain States Genetics Foundation/Mountain States Genetics Network

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**Statement of Peter Sybinsky, Ph.D.
Chief Executive Officer, Association of Maternal and Child Health Programs**

**Before the Secretary's Advisory Committee on Heritable Disorders and Genetic Diseases in
Newborns and Children
January 14, 2005**

Mr. Chairman and members of the Committee, I am Peter Sybinsky, CEO of the Association of Maternal and Child Health Programs. AMCHP is the national organization representing state and territorial public health leaders whose mission is to improve the health and well being of all women, children and families, including those with special health care needs. Our members administer statewide programs such as newborn screening, clinics for children with special health care needs, school-based health centers and a wide range of other vital health programs.

I would like to thank this Committee for your attention to this critical public health issue—newborn screening. Today, I would like to speak with you briefly on two specific issues:

1. The importance of addressing the entire newborn screening system, including follow-up and the key role of state family health programs, and
2. The need for adequate resources to support state newborn screening systems.

Testing newborns for metabolic disorders is essential to giving every child a strong start in life. However, effective newborn screening is more than just testing; it is an entire system that includes parent and doctor education, medical referrals and follow-up, and on-going financial support for a child's medical care.

Maternal and child health agencies help establish and direct newborn screening systems in every state. These agencies assure newborns are screened, provide standards for testing, notify parents of test results, refer parents to needed medical services, educate families and physicians of the implications of the test results, and conduct long-term follow-up with families to ensure they don't fall through the cracks. Once a child has been tested, there are still many activities to carry out to make sure those with a presumptive positive screen receive diagnostic testing from a qualified specialist and then get the services they need. In Tennessee, for example, the Maternal and Child Health program ensures that an infant with a presumptive positive test receives confirmatory testing, diagnosis and treatment, if necessary. That process includes written contact with the parent, the healthcare provider, a genetic center, endocrinologist or sickle cell center. The state program also arranges for any repeat specimens that may be necessary.

As the Committee moves forward with its work, AMCHP urges you to consider all aspects of state newborn screening systems, not only the actual blood spot testing but also the extensive follow-up activities by the health providers and the state maternal and child health programs.

The timeliness and accuracy of testing and follow-up services can make tremendous differences in the life of a newborn. However, we know from Kay Johnson's presentation at your previous meeting that these activities come with a price. According to the General Accounting Office, states spent over \$120 million on newborn screening in 2001. Laboratory costs accounted for 74 percent of states' expenditures, and only the remaining 26 percent paid for follow-up services. Many states rely significantly on the fees charged to families or hospitals to cover the testing and laboratory costs, while the Maternal and Child Health Block Grant pays for follow-up services and specialty care when it is not available through Medicaid, SCHIP, private insurance or other areas.

As newborn screening programs expand, someone will pay for the additional testing and follow-up. States continue to face budget limitations and expanding public health responsibilities. In this environment, it is difficult for states to maintain existing services, much less meet the needs of an expanded screening program. And it is unrealistic to expect families to pick up the additional cost of screening. In all likelihood states will be expected to pick up these costs.

As state budgets slowly recover from the economic downturn, new public health demands and rising healthcare costs, Federal support for programs like the block grant and Medicaid is even more crucial. AMCHP encourages the Committee to recommend that the Secretary support increased funding for the Maternal and Child Health Block Grant in the President's budget so that state maternal and child health programs can meet the increased needs of newborn screening programs. AMCHP also calls on the Committee to recommend full funding of Title XXVI of the Children's Health Act of 2000 and increased funding for CDC's efforts to help state newborn screening programs develop integrated data and surveillance systems. These systems allowing programs to link data to assist families to needed follow-up and treatment. In addition, adequate funding for Medicaid and SCHIP will be critical to the success of expanding newborn screening programs.

In conclusion, AMCHP strongly encourages this Committee to consider the real cost of newborn screening programs, including follow-up activities, and recommend adequate funding to link all families with the appropriate care.

The recommendations of this Committee can greatly strengthen state newborn screening systems and maximize the value of the additional screening tests. AMCHP thanks you for your efforts and hopes you will contact us for any assistance we can provide as you move forward in your work.

Thank you for the opportunity to provide input to this important effort.