

# State Development Grants for Newborn Screening Efforts and Infrastructure Development

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## Summary of the March 7, 2002 Meeting

Prepared for:

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## A. Introduction

In 1999, at the request of the Maternal and Child Health Bureau (MCHB) within the Health Resources and Services Administration (HRSA), the American Academy of Pediatrics (AAP) convened a Task Force on Newborn Screening to examine the many issues that have arisen around State newborn screening programs. After reviewing some of the challenges facing these programs, the Task Force made a recommendation to MCHB to provide States with grants that would help them stimulate development of newborn screening systems that are connected to the medical home and are integrated with other public health systems. These State planning grants, first awarded in 1999, aim to set a framework for establishing a genetic service infrastructure within States, and to create partnerships among State public health programs, primary care providers, the genetics community, and service consumers.

The purpose of the March 7, 2002 meeting, “State Development Grants for Newborn Screening and Infrastructure Efforts” was to bring together five States recently awarded grants to discuss the progress they have made thus far and the challenges they continue to face. Marie Mann, M.D., M.P.H., Deputy Chief of MCHB’s Genetic Services Branch, noted that the Branch hopes States will use newborn screening to build upon and link into other service programs, thereby facilitating long-term monitoring and continuity of genetic services. MCHB also hopes that in the third year of this initiative, States will begin the process for developing State genetics plans, and build on efforts to create a smooth interface between the public health system, community-based health care providers, consumers, and the genetics community.

Deborah Linzer, M.S., a Senior Public Health Analyst in the Genetic Services Branch, then went on to further describe the Branch and its activities. The mission of the Genetic Services Branch is to facilitate early identification of individuals with genetic conditions and integrate them into systems of care, and to increase the knowledge of genetic contributions to health and disease. The Branch views families as essential partners in this work, and has always encouraged States to keep families at the table in implementing any genetics policies or programs.

Many of the Branch’s current programs came about as a result of the recommendations offered by the Newborn Screening Task Force. The Task Force called for a national agenda on newborn screening programs, but found that there was little national oversight or consistent standards for treatment in State programs. The Task Force also found that States are ill-prepared for expanding their programs to include newer technologies. Following these

findings, the Task Force created a national framework for action, which recommends a systems approach to dealing with these issues, and emphasizes the need for any models for newborn screening and other genetic services to fit within States' existing infrastructure. Ms. Linzer noted that, in addition to offering State planning grants, the MCHB is further building on these recommendations by currently funding newborn screening implementation grants, and grants that allow States to develop models and materials to explore the clinical validity of new technologies in newborn screening.

There is additional funding for projects on sickle cell disease, Hemophilia Treatment Centers, comprehensive thalassemia centers, a newborn screening and genetics resource center, and a genetic evaluation needs project cosponsored by the March of Dimes. MCHB also has begun to conduct a series of genetics policy forums in conjunction with the Association for State and Territorial Health Officials (ASTHO) and the National Conference of State Legislatures (NCSL). Finally, the Agency is sponsoring a collaborative effort entitled "Genetics in Primary Care," which aims to provide in-depth training for faculty, students, and medical researchers on a variety of genetics issues related to primary care practice.

## **B. The National Newborn Screening and Genetics Resource Center**

Following Ms. Linzer's introduction to the Genetic Services Branch, Bradford Therrell, Ph.D., Director of the National Newborn Screening and Genetics Resource Center (NNSGRC), provided an overview of the Center. The NNSGRC is a cooperative agreement between MCHB and the University of Texas Health Science Center at San Antonio and serves to assist States and MCHB in outlining national policy, providing technical assistance for newborn screening and genetic services, and serving as an educational resource. One of the ways the Center fulfills this mission is by having a newborn screening technical assistance review team that offers help to States in enhancing their newborn screening programs. Dr. Therrell stated that the Center also has received funding recently to set up a similar review team in genetics that will assist States in developing their State genetics plan.

Dr. Therrell then presented the typical newborn screening workflow. The screening process begins shortly after a child is born, when identifying information is entered into the hospital accounting system. Patient information is transferred to the nursery and a newborn screening test is ordered, to be performed before the newborn is discharged from the hospital. In newborn screening, a blood specimen is collected from a heelstick, sent to a testing laboratory, and the prescribed tests (according to State law) are performed. Resulting information is

entered into a database, and test results are reported to the newborn's physician. A follow-up coordinator associated with the screening program is responsible for ensuring that any newborn whose results indicate the possibility of a disorder receives diagnostic follow-up. More tests and diagnosis from a subspecialist are required when screening results are positive. The workflow is similar in newborn hearing screening, which is being adopted and integrated in many States.

The feasibility of whether States can bring the data elements in newborn screening together and use them in some integrated fashion within the public health department is being addressed. Such "data warehousing" might allow for the capture of general demographic information by whichever program encountered the patient first, and could be accessed and expanded by more programs as the child ages (e.g., immunization registry). Dr. Therrell observed that newborn screening programs are good candidates for database sharing, and he presented examples of how many States have begun to combine data by bringing hearing screening results onto the same form as newborn screening. In showing a potential mechanism by which such data integration could occur, Dr. Therrell cautioned that several issues must be considered in the context of such database sharing. Some of these include: 1) whose information should be included, 2) how should consent to have and share information be obtained, 3) how can privacy be ensured, 4) can such information be shared between States when people move, and 5) when and how will the Health Insurance Portability and Accountability Act (HIPAA) apply.

Dr. Therrell then went on to identify other resources the NNSGRC can provide to States wanting to integrate genetic services into public health. These include:

- The NNSGRC's Advisory Committee on Genetics has as its objectives to provide technical assistance to States applying for planning grants, and to develop guidance for State genetics planning (which it has done in the form of a tool for integrating genetics into other public health divisions).
- A monograph put together with HRSA and the Council of Regional Networks for Genetic Services (CORN) was revised to become guidelines for integrating genetic services into public health. This guidance derives its constructs from MCHB and Institute of Medicine models, and brings in health care providers and genetics to a pyramid that demonstrates the overlap between types of services (direct health, population-based, enabling, and infrastructure building) and the core functions of assessment, policy development, and assurance.
- Genetics Education Materials (GEM) is a searchable database of online clinical materials and public health genetics policy documents.

- Finally, the NNSGRC Web site contains information and linkages that may be useful: <http://genes-r-us.uthscsa.edu>. Some State genetics plans and links to other State genetics plans are currently available on the site, and all States are encouraged to submit their plans to NNSGRC once they are completed.

### C. Title V Block Grant Program

Kathryn Peppe, R.N., M.S., formerly Chief of the Division of Family and Community Health Services at the Ohio Department of Health, followed Dr. Therrell's remarks by summarizing the involvement of Title V in the identification of children with special health care needs. Historically, maternal and child health has long been involved in issues of genetics and special needs, and through Title V, MCHB has received funding to support activities in this area. Title V also provides a mandate to States and the Federal government that assures the health of all women and children.

One of the critical components to the administration of Title V is the emphasis on active involvement of families. This level of involvement has steadily increased over the last decade. A survey by the Association of Maternal and Child Health Programs in 1992 revealed that families were involved in programs in only 21 States, while a later survey in 1998 revealed that this number had risen to 43 States. Family representatives were more likely to be involved in programmatic areas such as children with special health care needs, early intervention, and managed care than in areas such as HIV/AIDS or prenatal care. The nature of work offered by family representatives included budgetary decision-making, statewide needs assessment, serving on advisory committees, and parent support and mentoring. Ms. Peppe stressed that it is important that States consider using families in all areas of programming, because they can have a positive impact on planning, implementation, and outcomes.

### D. Overview of the Association of State and Territorial Health Officials (ASTHO)

Another resource for States planning newborn screening efforts is the Association of State and Territorial Health Officials (ASTHO). Amy Klein, M.P.H., Director of Genetics at ASTHO, provided an overview of the organization and the history of its genetics program.

In 1998, the Centers for Disease Control and Prevention provided ASTHO with funding to begin its genetics program, and a year later, ASTHO's Genetics and Public Health Workgroup was formed. In 2000, ASTHO formed a Genetics Advisory Committee (GAC) with members from State departments of health and affiliate members representing other genetics and public

health agencies and organizations. Working from a framework document devised by the Genetics and Public Health Workgroup, the GAC refined this document and used it as the basis for ASTHO's Public Health Genetics Policy Statement. This statement addresses specific issues such as privacy and confidentiality, genetic discrimination, informed consent, screening, public health workforce competencies, and eugenics.

In addition to the Genetics Policy Statement, ASTHO has also been working on a document entitled "Framework for Integrating Genetics into Public Health." This framework addresses core functions such as assessment, policy development, and assurance, and is predicated upon the following beliefs:

- Genetics will become a fundamental component of the policy and practice roles of public health agencies by 2010.
- Breakthroughs in human genetics provide great promise for improving the health of the public, but there are significant policy implications and resource needs.
- Genetics will offer many opportunities for public and private collaboration, but State health agencies will bear the ultimate responsibility for ensuring that genetic information is integrated into the basic scientific and technical knowledge of public health appropriately.

The framework goes on to identify 10 essential services public health departments should take into account when integrating genetics into their services and programs.

Ms. Klein also highlighted several other initiatives being undertaken by ASTHO at this time. Beginning in 2002, ASTHO has, along with HRSA and NCSL, launched a series of genetic policy forums designed to educate legislators and senior State public health officials about implications of new genetic science on public health. For these forums, ASTHO has developed a series of policy briefs, which currently are available on their Web site ([www.astho.org](http://www.astho.org)). Finally, ASTHO is working on a Genomics and Public Health Toolkit to improve genomics capacity in State and local health agencies by assisting them with integrating genomics into public health practice and policy.

## **E. State Presentations**

After having the opportunity to learn more about what progress was being made by organizations in the area of genetics and public health integration, the five States in attendance

were given the opportunity to report on their progress and challenges so far in implementing newborn screening efforts. What follows is a summary of their comments.

## 1. Connecticut

Representatives from the State of Connecticut discussed how they are working toward meeting the four objectives of their planning grant: to conduct a statewide genetics needs assessment, develop partnerships, assess data capacity, and finally, develop a State genetics plan. The State began preparing to meet these objectives by identifying key stakeholders among different provider groups, including primary care physicians, specialists in neonatology, perinatology and genetics, medical organizations, families, and offices within the State Department of Public Health and other State agencies.

A recent State needs assessment was conducted, which identified referral patterns in 30 Connecticut hospitals. Among these hospitals, it was found that eight offered genetic counseling and laboratory services on-site. In the remaining 22 facilities, services were provided through contracted outreach or referral. The State will perform a broader needs assessment to identify future directions and gaps in programs, and to prioritize needs around the role of genetics in public health. Overall, the needs assessment will address current and future genetic issues, including education and data integration. Consumer input will be sought throughout this process.

Connecticut's goals for data integration are to assure early identification and referral of children with special needs to appropriate service providers, develop partnerships among internal and external State agencies and private entities to deliver coordinated genetic services, and increase coalition building among all stakeholders who will be impacted by a newly developed State genetics plan. The State has already identified data sources to feed into the integration effort, including newborn screening programs, immunizations, vital records, and lead poisoning prevention. The challenge they now encounter is answering the following questions:

- What do we want linked (data elements and systems)?
- What systems do we want integrated?
- Does linkage give the same product as integration?

The State is also evaluating its progress on a number of Title V performance measures which will help determine how infrastructure development and the State Genetics Plan can help it better measure and achieve these objectives.

## 2. Minnesota

Representatives from the State of Minnesota emphasized that while their process and objectives for their planning grant is similar to Connecticut's, they have also made a significant effort to involve families in every step of this process so as to develop an infrastructure that will be most useful to families. Minnesota has found through its initial needs assessment that families often do not know for which services they are eligible, and how to access them, and thus the State is trying to bring in two parents to serve on every working group they form during this process. They noted that while the State is rich in the number of services it provides, these are often fragmented and therefore inaccessible to families.

Prior to receiving the grant, Minnesota's newborn metabolic and hearing screening programs were already in the process of integrating, but the State hopes to use the grant to extend these efforts to incorporate early identification programs and primary and specialty care providers. The desired outcomes of this effort would be improved service delivery of newborn screening, enhancement of data capacity, and ongoing monitoring of genetic technology in newborn screening.

While the work groups established around the needs assessment and data integration issues have been committed to these efforts, Minnesota has faced some challenges in staffing these initiatives. They have also been trying to find ways to allay the public fears about how these data will be used.

## 3. Nebraska

While the State of Nebraska has a strong emphasis on using its grant to improve newborn screening, the State, like Minnesota, is also trying to broaden the grant's application to children with special health care needs (CSHCN). In its first grant year, the State has worked to form a State Genetics Advisory Committee with a broad representation of providers, consumers, and others, and this group has developed an operational definition of CSHCN with genetic conditions. The Committee has also formed a charter and established a series of work

groups around the following issues: policy, data infrastructure, personnel resources, financing, and legislation.

After the work groups convened via telephone conferences and identified perceived barriers in current services and ways to make improvements in these, they created surveys around collaboration, insurance, databases, physicians, and parents of recently screened newborns. It is expected that the results of these surveys will provide further recommendations for improvements in services. The State has also successfully obtained information from other sources—including genetic counselors, the NNSGRC, literature reviews, and the Internet—to provide additional comments to the work groups.

Nebraska's concerns at this juncture have been related to the level of consumer involvement in this process, and the lack of broad minority and geographic representation. The State also expressed the need to have more specific guidelines for issues surrounding CSHCN, using tandem mass spectrometry, and Federal and State statutes and regulations (e.g., HIPAA). Nevertheless, the Committee is moving forward, and will meet this spring to develop a prioritized list of needs incorporating feedback from the work groups.

#### 4. North Carolina

As a recent beneficiary of a State planning grant, North Carolina is just beginning the process of putting disparate pieces into a coherent strategy for creating a statewide comprehensive genomics plan and assessing ways to integrate databases to better serve CSHCN. The State is hopeful that recent announcements from three major universities to begin genomics initiatives will bring further success to this process.

Thus far, North Carolina has identified a State team representing a cross-section of agencies and services, including public health departments, the CSHCN program, the Early Intervention Program, the State's center for health statistics, and epidemiology center. The State hopes to capitalize on the strong relationships it has with the State pediatric society, North Carolina medical society, March of Dimes, and the network of regional genetics centers to further its work on meeting the objectives of the grant.

North Carolina has already experienced rapid implementation of universal newborn hearing screening and universal screening using tandem mass spectrometry. The State has a positive relationship with a State senator who supports electronic birth certificate data; however, the

State is most concerned about a \$1 billion budget shortfall and the ensuing cuts that will occur in State programs in the late spring that might affect its progress in this area.

## 5. Tennessee

Representatives from Tennessee noted that one advantage to their newborn screening program is that they already have a really good follow-up system in place for infants identified through the screening process. Thus, the focus of their grant is to coordinate this good system with other early identification programs, and find ways to manage and share data across these programs. To do this, they have chosen to further expand the newborn screening program by adding hearing screening and other conditions, and to begin promoting coordination of services by profiling some of the data to emerge from the newborn screening program. Other immediate objectives for the State include:

- Distributing audience appropriate literature;
- Developing more effective educational tools for providers;
- Coordinating newborn screening follow-up with medical homes;
- Promoting knowledge about genetic services;
- Engaging providers in conducting needs assessments; and
- Increasing consumer participation.

Tennessee has begun to address its coordination goals by strengthening its already existing Genetics Advisory Committee, to include more members and form genetic subcommittees. These subcommittees deal with topical issues such as adding conditions to the newborn screening program, ethics, professional education, and consumer involvement.

The State has also been engaged in a collaborative effort called Children's Information Tennessee, to put together data information and condense data "silos". A memorandum of understanding around this effort has already been signed off on by members of health, children's services, education, and human services departments. Information is accessible through a Web interface, and the ability of providers to view the information requires a release signature from a parent/guardian.

## F. Breakout Sessions

Following presentations by the States, the participants assembled into a series of breakout sessions designed to discuss issues of importance to their newborn screening and general genetics and public health activities. Each session—which covered the topics of informatics, genetics planning, partnership, and evaluation—allowed States the opportunity to communicate further their efforts and challenges in each of these four areas.

### 1. Informatics

The informatics breakout session began with a series of ad hoc observations regarding the States' earlier presentations. Barry Nangle, Ph.D., Director of Utah's Office of Vital Records and Statistics, noted that too often parents are asked to fill out forms containing the same information many different times. One goal of the States' grant enterprise should be to form a seamless system of genetic services from screening to community-based services that reduces redundant data collection. Edward Gotlieb, M.D., Chair of the AAP's Task Force on Medical Informatics, stated that so far the discussion around integration has not involved the private sector; States should not talk about achieving integrated systems while neglecting a whole section of the medical community.

Participants were particularly keen on addressing this latter issue, and raised a number of points in relation to involving private practitioners in their data integration and genetics planning efforts:

- They observed that the key to giving providers data is finding out what format would maximize the utility of that data for them, especially given time constraints. Most physicians don't want to interrupt their already busy office flow to fill out more forms or spend time trying to interpret data.
- Participants also stressed the need to have data available at the time infants will be visiting the pediatrician (e.g., their two-week check-up). It is disruptive to both families and physicians if they must reschedule visits to discuss screening results.
- Another participant challenged the notion that knowledge flows down to the practitioners. Instead, States should consider how to create databases of information that are interactive and in which information flows in many directions.

The participants also agreed that it is crucial to examine the needs of the end user to determine what information will ultimately benefit that person. Electronic medical records should not

necessarily be considered a panacea. As one State representative noted, paper records are portable, inexpensive, and flexible to process; any IT solution must carry those same standards.

The breakout session concluded with participants expressing some of their concerns about challenges they are facing in developing information technology solutions in their States. Tennessee noted that State rules and regulations can force practices to evolve; such is the case with their laws on hearing screening, which do not mandate the screening, but which require physicians to report results if they do conduct this screening. Another State noted that it is challenged by consumers' concerns about data collection and confidentiality, and is still trying to determine who gets access to this information. Finally, one participant noted that relying solely on pediatricians to access information neglects the large number of children who do not have a medical home.

## 2. Genetics Planning

The genetics planning breakout session, facilitated by Brad Therrell and Kathryn Peppe, attempted to address some of the processes necessary for carrying out the objectives of the planning grants, and to identify long-term goals of these processes. The group started by noting the barriers to planning, namely that their projects were sometimes deemed as lower priorities compared to other State health initiatives, and the limited length of the grants do not help them to plan for sustainability. The participants also observed that genetics is defined differently across States, but they agreed that States need to stress how genetics impacts everyone.

Participants noted that the majority of their State genetics programs are newborn screening, and that there are multiple issues that have been under-addressed. They cited specifically:

- States need to identify and perform meaningful comprehensive program evaluations that go beyond merely counting the number of tests performed.
- States need to consider how their program results compare to *Healthy People 2010* performance measures and outcomes.
- Several participants questioned the time limit of a State genetics plan, since genetics feels like a constantly moving target. There were suggestions either for setting expiration dates for plans (as is done in Ohio), building in periodic review phases and updates, and conducting periodic needs assessments, with input from the advisory committees.
- Making the planning process inclusive of a variety of professionals and consumers was also a concern. Some participants recommended developing train-the-trainer

consumer programs, and emphasizing professional development as ways to bring about meaningful participation.

- Finally, States questioned how private industry can influence the planning process. They felt that more thought should be put into ways of cultivating public-private interfaces, especially given how the private sector can provide financial or human resources.

### 3. Partnerships

The partnership breakout session began with facilitators James Lustig, M.D., Vice President of Medical Affairs at Toledo Children's Hospital, Patti Hackett, M.Ed., Executive Team Leader, and Glen Gallivan, Turf Writer and Associate Director at Hackett Solutions, asking participants what they wanted to achieve in the session. The participants established as their main goals trying to understand how genetic information is translated between professionals, policymakers, and consumers, and the discussion of practical issues related to primary care providers of CSHCN.

When asked how States were getting the participation of families and consumers, several replied that meaningful compensation played a role. Some participants noted that they found it difficult to provide financial compensation. A participant from the State of Tennessee noted that the State contracts with parents as consultants. All the States agreed that more information was needed on successful family compensation, to include rates, expectations, and roles. One participant suggested the Ryan White Foundation as a possible resource.

The group then listed some of the barriers to participation of families, including transportation, costs of day care and time off of work, and burnout. Nebraska noted a barrier in getting Tribes' participation because of their inability to send members off the reservations and into genetic communities. All of the States agreed that to maximize participation from families, consumers, and representatives from family and youth leadership organizations, they need to clearly articulate what an organization/committee wants to achieve, and the expected roles and responsibilities of those who participate.

The conversation then shifted to some of the practical problems primary care physicians encounter when treating CSHCN, including limited time with patients, paper records, unintegrated data systems, and increased pressure for documentation. Additionally, since many diseases have specialty clinics associated with them, information does not always flow from these to the primary care doctor. Other challenges noted by the group included:

- There is a need for information provided to consumers to be written in person-first language, rather than focus solely on the diagnosis/condition (e.g., neuromuscular afflicted versus a child with neuromuscular disease).
- Finding ways to assure that services are designed in response to the needs of families, children, and youth, thereby increasing positive outcomes and accountability. The goal is to strengthen coordinated services without duplicative components that tax families' energy and/or waste their time.
- There is a need to share medical documentation with families and providers to expedite paperwork needed to qualify for programs.
- There is also a need to think holistically and lay the ground work for better coordinated services and increased patient/family education in decision-making to benefit the process of transition for CSHCN from pediatric to adult services.
- Emphasis should be placed on supporting the skill-building of families and assuring that information requested of families is coordinated and shared among agencies, when possible, to avoid duplication of effort. Families should also be included as partners in the development and design of integrated information data systems.

All of these challenges, the participants concluded, could begin to be dealt with by fostering collaborative partnerships across agencies, with families, consumers, and representatives from family and youth leadership organizations, and with providers.

#### 4. Evaluation

The evaluation breakout session began with a brief presentation on the goals of the evaluation process by Nicole Fehrenbach, M.P.P., Senior Research and Evaluation Associate at the Center for Innovation. Ms. Fehrenbach explained that evaluation is intended to make judgements about the effectiveness of a program and inform future program decisions, and that in no way should it occur solely at the end of a program. Within evaluation, standards have been established around utility, propriety, feasibility, and accuracy. In addition to these standards, there are both external and internal goals to be satisfied.

She recommended that with the newborn screening planning grants, the evaluation approach should center on the following: defining the program, monitoring it, process measures, goal and objective clarification, discrepancy analysis, and needs assessments. To approach the evaluation in this way, Ms. Fehrenbach suggested that States make evaluation someone's job specifically, budget 5-10% for the process, commit to careful documentation, incorporate multiple perspectives, and ask reflective questions about the project's intent, value, and methodology.

While States agreed with the goals of evaluation, they expressed concern that given the limited amount of money offered in grants, they would be unable to commit the types of resources to performing an evaluation. There was especially concern about whether the people who applied for the grant should be the same ones performing the evaluation. One participant stressed, however, that stakeholder-based evaluation is a new philosophy that is slowly taking hold, and that one can have a valid internal evaluation after defining, with outside stakeholders, a series of “good” indicators.

## G. Charting the Course

Once participants had the opportunity to convene and discuss specific issues of importance to their progress, Alan Hinman, M.D., M.P.H., Principal Investigator for the Center for Innovation, attempted to reiterate the major themes of the day and provide a summary of progress made in newborn screening. He began by outlining the process of development immunization registries underwent a few years back, and how these can model lessons learned for newborn screening.

In a major report on immunization registries issued by the National Vaccine Advisory Committee, the Committee outlined the main issues facing registries, namely to ensure appropriate privacy protections, to ensure participation of all immunization providers and recipients, to ensure appropriate functioning of registries, and to secure sustainable funding for them. Immunization registries have demonstrated their usefulness in many ways, including by increasing coverage, generating official records, reducing missed opportunities, and providing vaccine inventory management. However, there are some remaining challenges to these registries. In addition to the need to maintain their initial momentum, immunization registries have yet to link to other registries and information systems, nor has there been an emergence of 3-5 standard, reliable software packages with which they can be implemented.

Newborn screening can learn a lot from immunization registries, because it faces many of the same issues, and newborn screening systems continue to encounter problems in areas of follow-up and linking and integrating systems. Immunization registries also are similar to newborn screening systems in these ways:

- They must be established at, or near, birth;
- They are population-based;
- They support clinical decision-making at the point of service; and

- They provide a summary of public health information, and means of assuring follow-up and completion of recommended action.

HRSA recognized that these problems will only occur more frequently as more screening is done in the future, and thus created these planning grants as a way to help States prepare for this. Linkage and integration are important foci of these grants because they focus on allowing a smooth exchange of information, and relate ultimately to the end user, not the machinery with which they operate.

After providing several examples of why it is crucial to have information systems to assure follow-up, Dr. Hinman reiterated the cross-cutting themes found in the State presentations. These included:

- States are in varying stages of progress with their grants.
- The role of genetics in public health needs to be clearly defined to expand the thinking around traditional newborn screening programs to a broader consideration of genetics.
- States need to take the separate components of existing and new programs and make them into a coherent whole.
- An active, broadly represented advisory group is essential to the success of programs.
- Partnerships should be cultivated—with families, other health care providers, policymakers, insurance companies, and other industries.
- States are still dealing with how to handle issues of privacy, confidentiality, and discrimination, and are challenged to define the boundaries for screening (especially in policymaking).
- States recognize the necessity to involve providers, families, and communities at all stages, and to find ways to have information systems communicate effectively.
- Finally, States realize that lack of financial and human resources can impede their progress in establishing their State genetics plan.

Lastly, Dr. Hinman offered the following recommendations to participants in continuing their progress on developing their newborn screening information systems and setting up a genetic services infrastructure in their States:

- Keep an eye on the goal—improved health through improved information and services.

- Assure broad participation in developing plans and implementing programs.
- Advocate for integration.
- Try to have early successes.
- Work towards standards.
- Learn from other programs.
- Keep in touch with other States and learn from each other.

Appendix A: List of Participants

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Maternal and Child Health Bureau

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Appendix B:            Agenda

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U.S. Department of Health and Human Services  
Health Resources and Services Administration  
Maternal and Child Health Bureau

# State Development Grants for Newborn Screening Efforts and Infrastructure Development

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*Bethesda Marriott  
Bethesda, Maryland  
March 7, 2002*

## Agenda

**States:** Connecticut, Minnesota, Nebraska, North Carolina, Tennessee

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|--------------------|--|
| 7:30 – 8:00 a.m.   | <b>Continental Breakfast</b>   |
| 8:00 – 8:15 a.m.   | <b>Welcome and Introduction</b><br>Marie Mann, Deputy Chief, Genetic Services Branch   |
| 8:15 – 8:45 a.m.   | <b>Genetic Services Branch: Mission, Programs &amp; Activities</b><br>Deborah Linzer, Senior Public Health Analyst, Genetic Services Branch  |
| 8:45 – 9:30 a.m.   | <b>Genetics and Public Health: Integrating/Linking Information Systems and Developing Meaningful Program Infrastructure</b><br>Brad L. Therrell, Director, National Newborn Screening and Genetics Resource Center |
| 9:30 – 9:45 a.m.   | <b>Welcome, Federal Title V Block Grant Program</b><br>Kathryn Peppe, Independent Consultant, Formerly Chief, Division of Family and Community Health Services, Ohio Department of Health                          |
| 9:45 – 10:00 a.m.  | <b>The Association of State and Territorial Health Officials (ASTHO) Overview</b><br>Amy Klein, Director, Genetics Department, ASTHO   |
| 10:00 – 10:05 a.m. | <b>Introduction of Facilitators, Housekeeping</b><br>Kathy Peppe<br>Vivian Ota Wang, Professor, Counseling Psychology, Arizona State University  |

10:05 – 10:20 a.m.	<b>Break</b>
10:20 a.m. – 12:00 p.m.	<b>State Presentations</b> <ul style="list-style-type: none"> <li>• Connecticut</li> <li>• Minnesota</li> <li>• Nebraska</li> <li>• North Carolina</li> <li>• Tennessee</li> </ul>
12:15 – 1:15 p.m.	<b>Lunch</b>
1:15 – 2:15 p.m.	<b>Breakout Session 1a</b>  <b>Informatics</b> Barry Nangle, Director, Office of Vital Records and Statistics, Utah Department of Health Ed Gotlieb, The Pediatric Center  <b>Breakout Session 1b</b>  <b>Genetics Planning</b> Brad Therrell Kathy Peppe
2:15 – 3:15 p.m.	<b>Breakout Session 2a</b>  <b>Partnerships</b> Jim Lustig, Vice President of Medical Affairs, Toledo Children's Hospital Patti Hackett, Executive Team Leader, Hackett Solutions Glen H. Gallivan, Turf Writer, Beyer Speed Analyst, Hackett Solutions  <b>Breakout Session 2b</b>  <b>Evaluation</b> <i>Taskforce for Child Survival and Development</i> <i>Center for Innovation</i> Dave Ross, Director Alan Hinman, Principal Investigator Nicole Fehrenbach, Senior Research and Evaluation Associate Ellen Wild, Director of Programs
3:15 – 3:30 p.m.	<b>Break</b>

3:30 – 4:30 p.m.

**State Teams Revisit Work Plans/Matrices**

Facilitator: Vivian Ota Wang

Technical Advisors: Dave Ross  
Alan Hinman  
Kathy Peppe

4:30 – 5:30 p.m.

**Charting the Course: Discussion of State Presentations**

Alan Hinman

5:30 p.m.

**Adjourn**