

Translating Recommendations into Action

Within the funds provided [for 1999-2000], the Committee urges the availability and accessibility of newborn screening services to apply public health recommendations for expansion of effective strategies. HRSA, in collaboration with the Centers for Disease Control and Prevention (CDC) and the National Institutes of Health (NIH), is encouraged to develop and implement a strategy for evaluating and expanding newborn screening programs, pilot demonstration projects, and the use of contemporary public health recommendations on specific conditions, such as cystic fibrosis and the fragile X syndrome. If implemented, the Committee directs that tangible steps be taken to protect patient privacy and to avert discrimination based upon information derived from the screenings.

— Senate Finance Committee

Objectives

- To identify strategies for implementing recent recommendations to achieve Healthy People 2010 objectives for newborn screening and genetics.

Outcomes

- Identify for HRSA and MCHB priority areas in the translation of recommendations into action.
- Identify recommendations for the use of SPRANS funds in the translation of recommendations into action.

Post Meeting Products

- MCHB Action Plan for the translation of recommendations into action.

Participants/Observers

- Leaders in the field from State government, Federal government, MCHB-funded projects, consumer organizations, and the genetics profession.

Translating Recommendations into Action

Agenda

- 9:30 AM - 10:00 AM** **Welcome & Introductions**
Claude Earl Fox, MD, MPH
Merle McPherson, MD, MPH
Facilitator: John Reiss, PhD
- 10:00 AM - 10:20 AM** **The Human Genome Project and Medical Practice**
Ed McCabe, MD, PhD
- 10:20 AM - 10:40 AM** **State Genetic Resources and Services:
Findings from a national survey**
Joann Lindenmayer, DVM, MPH
- 10:40 AM - 11:00 AM** **An Ethnocultural and Geographical
Approach Toward Genetic Literacy**
Kathleen Rand Reed
- 11:00 AM - 11:15 AM** **Charge to the Group**
Michele Lloyd-Puryear, MD, PhD
- 11:15 AM - 12:30 PM** **Consumers as a resource**
Mary Davidson, Alliance of Genetic Support Groups
Donna Olsen, Family Voices
- 12:30 PM - 3:30 PM** **Small Group Discussions**
Issues, Forces & Factors
Facilitator: Ed McCabe; Resource: Brad Therrell
Assuring Access & Quality
Facilitator: Bob Fineman; Resource: Kay Johnson
Data & Surveillance
Facilitator: Paul Nannis; Resource: JoAnn Lindenmeyer
Literacy & Training
Facilitator: Joe McInerney; Resource: Kathleen Rand Reed
**Partnerships for Successful Outcomes for Families
and Professionals**
Facilitator: Mary Davidson; Resource: Donna Olsen
- 3:30 PM - 5:00 PM** **Group Presentations & Discussion**
Michele Lloyd-Puryear, MD, PhD
Merle McPherson, MD, MPH
Facilitator: Kay Johnson

What Recommendations are Before Us?

The Newborn Screening Task Force outlined a national agenda for strengthening each state newborn screening system that calls for:

1. Defined public health responsibilities for Federal and State public health agencies;
2. Model regulations to guide implementation of State newborn screening systems;
3. Minimum standards for newborn screening activities (e.g., sample collection, laboratory quality, sample storage, and information systems);
4. Model follow-up, diagnosis and treatment guidelines and protocols for health professionals and other participants in the newborn screening system;
5. Designs for systems of care that fit the national framework for CSHCN (birth to adulthood);
6. Effective tools and strategies to inform families and the general public; and
7. Demonstration projects to evaluate technology, quality assurance, and outcomes.

The Task Force made further recommendations to address specific concerns and identified needs for program and policy development in four key areas: public health infrastructure, professional and consumer involvement, surveillance and research, and adequate funding.

Enhancing State Genetic Programs: Summary of Recommendations for Public Health Agencies

(Based on the results of a survey conducted by the Council of State and Territorial Epidemiologists)

Coordination and collaboration

- Develop a comprehensive approach to planning for genetic science.
- Coordinate existing categorical programs in MCH, chronic disease, and environmental and occupational health with new initiatives in genetics.
- Identify public and private partners to ensure that effective strategies for developing and sustaining public health capacity in genetics is achieved.

Categorical program linkages

- Develop funding capacity for new areas in genetic practice, particularly those in chronic disease and occupational and environmental health that may not have categorical funding for genetic activities;
- Integrate existing MCH resources and expertise on genetic science into existing chronic disease and occupational and environmental health programs;
- Promote effective communication and coordination among these areas and between public, private, academic, and consumer organizations.

Needs assessment and planning

- Develop the capacity for and conduct early needs assessments;
- Plan for policy and program development as new genetic information regarding specific disease areas and prevention measures becomes available.

Laboratory and testing policy

- Consider the need for State regulation of genetic testing in private laboratories;
- Determine appropriate policy to prevent the undesired use of genetic test results;
- Analyze new genetic tests to determine clinical utility and applicability to public health programs.

Staff capacity

- Employ a Genetic Coordinator to promote the effective use of relevant genetic information for public health;
- Improve State Department of Health staff knowledge of genetic information and technology;
- Take advantage of available training opportunities to develop State capacity in genetic information and technology.

Welcome by C. Earl Fox, MD, MPH, Administrator, HRSA

Introduction

It is a pleasure to welcome you all to this strategic planning meeting where important decisions will be made about how we together can meet the challenges and opportunities posed by the recent recommendations of the Newborn Screening Task Force.

Because of our mission and abiding commitment to providing and improving access to care for the nation's most underserved people, HRSA is uniquely positioned to play a leadership role in improving the quality of genetics services for American families who need these programs most. Long before there was a Human Genome Project and years before human genetics was prominent on the nation's radar screen, the Federal government's Maternal and Child Health program was leading the way in this effort.

MCH helped start newborn screening programs in every State and has given funding to every State for genetics services programs. Today, HRSA's Maternal and Child Health Bureau [MCHB] continues to carve out its leadership role in this key area. MCHB is working hard to: strengthen the current public health infrastructure to support proven genetics services for at-risk families; ensure the effectiveness of local newborn screening programs; and develop health policies to support and guide State and local programs.

Today's meeting agenda is critical because we are talking about the future of genetics services—about what it's going to take from all of us to move this effort forward into the new century.

Translating Recommendations into Action

While genetics research activities, particularly the Human Genome project, have enjoyed great visibility and generated considerable public interest, much less attention has been paid to how we can take advances in genetic resources and services and translate them into viable services that make a difference for children and families at the local level.

The Newborn Screening Task Force recommendations you consider today offer many opportunities to take us to another level. Certainly, they provoke the right questions: Do we have the infrastructure to adequately support quality newborn screening programs? Have we sufficiently educated the public about the benefits of newborn screening? Have we built in the necessary systems for data collection and analysis? Have we put in place adequate financing mechanisms to support comprehensive care? As you consider these questions, I urge you to put the emphasis on how best to integrate genetics services into existing programs, not on building new separate, free-standing entities.

Meeting One

MCHB's State planning grants work well for this very reason. They underscore the need for States to integrate newborn screening programs with other early identification programs, such as birth defects surveillance programs, immunization registries, and so forth. These planning grants will pave the way to create a larger role for maternal and child health programs in genetics policy. They will further our ability to do surveillance, to look critically at health services data, and to monitor systems and outcomes better.

Looking Ahead

At HRSA, we want to assure equity and access to genetic services in order to maximize benefits and minimize risks to all patients, families, and their communities.

We're finalizing a Memorandum of Understanding with NIH, the Agency for Health Care Policy and Research, and CDC that will serve as a mechanism for sharing information and expertise in genetic research, services, education, and policy. Through this MOU, we will collaborate and consult each other as we design and implement programs and activities relating to genetic information.

HRSA also supports the Alliance of Genetic Support Groups that builds partnerships among consumers and professionals to promote better health care for people with genetic conditions. This group is active in every State, and has available consumer indicators of quality genetic services in every State.

Looking ahead, our bottom line is clear: we want to support programs that incorporate genetics knowledge into existing public health systems. We want newborn screening programs that give all infants the same access to testing that can prevent impairment and disability. We want to ensure that all the new technologies are used appropriately and in a culturally sensitive manner.

I want to commend you all for your willingness to come here and address these tough issues. I look forward to sharing in your deliberations.

★ ★ ★

Introductory Remarks

Merle McPherson, MD, Director, Division of Services for Children with Special Health Needs, MCHB-HRSA

Let's begin with a review of historical efforts to create a community of services. Many people have been at it for a long time. The approach was to change from an isolated medical model to a family-focused, community-centered model in tune with the consumer movement.

- In 1988, Surgeon General C. Everett Koop introduced a National Agenda for CSHCN that has been endorsed by more than 70 professional and voluntary organizations.
- In 1989, Federal legislation [OBRA '89] set out requirements for family-centered, community-based, coordinated systems of care and services for CSHCN.
- Healthy People 2000 called for an increase in the proportion of states that have such service systems.
- In the context of the Government Performance and Results Act [GPRA] of 1994 and reflecting a growing movement toward results-based accountability in government and other health programs, the Federal MCHB set out a set of core national performance measures for Title V MCH Block Grant programs and called for States to set additional measures.

Healthy People 2010 does not have specific, detailed objectives for CSHCN; however, a related process provides an opportunity to move forward on full implementation of systems of care. This effort will provide a ten-year focus on achieving and measuring success for all CSHCN. During this ten-year window, the aim is for universal coverage and adequate systems of care. The process for achieving and measuring success is beginning now.

In partnership with the March of Dimes, MCHB has convened 6 workgroups. Each will focus on one of the performance measures from the National Agenda and create a ten-year plan. The group includes representatives of key constituencies with concerns about CSHCN. The combined ten-year action plan will form the basis for a monograph on CSHCN to accompany Healthy People 2010.

The time frame is:

- October 1999 – work groups established
- December 1999 – co-chairs meet in DC
- January 2000 – launch of Healthy People 2010
- November 1999 – June 2000 work groups meet
- June 2000 – action plans finalized
- October 2000 – monograph completed
- February-March 2001 – CSHCN national meeting held.

While the timetable is much longer for the challenge of incorporating genetic medicine and technology into practice and service, the genetic community should be a part of this effort. Children affected by genetic conditions are a subgroup of CSHCN and need the same system, coverage, and supports as children with special needs whose conditions have may non-genetic origins. In this context, it is important that the focus not be narrowed down to only a screening program. Medical homes, early intervention, involvement of families, and support services are essential components of a system of care for CSHCN.

Reflections on futurist viewpoints regarding health care:

- Managed care is not the solution some people thought it would be. We must decide how to modify and move forward and how to better manage health care without using managed care;
- The competitor is not a hospital in next county, it is e-commerce and the empowerment of consumers through the Internet. Links to consumers have been the focus in efforts to improve systems of care for CSHCN;
- Cultural and demographic changes are a major force. For CSHCN, cultural competency work is already underway;
- Scientific and technological advances are fast-paced and enormous. One of the key areas for scientific and technological change is genetic information and technology. The impact of genetic science on public health and health care delivery systems will be significant. The challenge is to ensure that these advances help children and are available on an equitable basis.

Environmental Context for Genetic Services Branch Activities, Dr. Michele Puryear, Chief, Genetic Services Branch

Strategic planning requires an assessment of the internal and external environment of an organization. To determine our role in translating recommendations into action, we have asked: What is the environmental context for the activities of the genetic services program at MCHB-HRSA? In recent years, five dramatic changes have occurred that affect genetic services.

Advances in Science

The Human Genome Project is fulfilling its promise to sequence and analyze genetic material. Human DNA sequencing is entering a decisive phase. The implications for biomedical science, medical practice, and public health decisions are enormous. The impact of this new scientific information will be felt in five key arenas.

- The Human Genome Project provides the basis for understanding variations in risk among individuals for medically important and genetically complex human disease. This new knowledge takes on even greater significance in the context of the changing demographics of our country, of concepts and misperceptions of race and ethnicity, and of an understanding of cultural differences in approaches to health.
- The resulting ethical, legal, and social questions also must be addressed. One challenge is to balance the need both to protect population health and to respect individual rights. The ethical questions related to screening children for additional conditions are another example of the type of issues that must be managed through public discussion and public policy.
- The combination of advances in genetic knowledge and rapidly evolving technology create the potential for using new tests for additional conditions, different screening methods for previously accepted conditions, and alternatives that lead to different levels of efficiency and cost-benefit. State policy makers will face growing dilemmas when making decisions about regulation of new tests and testing modalities.

Innovations in Information Technology

The capability to exchange information and the tools to analyze information are rapidly expanding. With bioinformatics, it is plausible that reliable prognostic information will allow individuals to make informed decisions and choices and health professionals to assist in the design of targeted preventive strategies. But while the expansion of information technology will allow greater communication and a deeper understanding of science, this new technology also brings many social, ethical and legal issues.

Health Care Delivery and Financing

The increased use of managed care arrangements has changed the mechanisms for both delivery and financing of health services. State newborn screening and related programs must adapt in response to this trend. In addition, because children are now more likely to have publicly-subsidized health coverage (through Medicaid and CHIP plans) and less likely to have employer-based coverage, States play a greater role in assuring the adequacy of child health benefits. In addition, States' oversight of health plans has increased, with virtually all having adopted laws or regulations related to quality or consumer protection in the new health system.

Public Health Agency Roles

As delivery and financing mechanisms have shifted, so have public health agency roles. Compared to the 1970s and 1980s, State and local health agencies are less likely to provide personal health services and more likely to contract with providers and health plans for delivery of services. However, public health agencies' responsibilities in assessment, assurance, and policy development remain. The Centers for Disease Control and Prevention [CDC] has an initiative underway to link public health and genetic activities. For MCHB and the genetic services program, new approaches and mechanisms in newborn screening systems and related early identification program must be developed.

Shifts in the Federal-State Division of Responsibility

Starting with the creation of the Maternal and Child Health Block Grant (1981) and continuing through the Government Performance and Results Act [GPRA, 1995], Personal Responsibility and Work Opportunities Act [PROWA, 1996] and Balanced Budget Act [BBA, 1997] that created the State Children's Health Insurance Program [SCHIP], the division of responsibility and level of accountability between Federal and State health agencies has been in flux. Today, State maternal and child health programs have greater flexibility and, at the same time, greater accountability.

★ ★ ★

Suggested Action Strategies

Outcomes from Meeting One were intended to be priority areas for action and recommendations for use of SPRANS funds. The participants concluded that MCHB should take action and support activities as follows:

1. Integrate genetic information and technology into MCHB activities across the lifespan of families with activities.
 - a. Centered in the Genetic Services Branch.
 - b. Part of: Perinatal Division; Division of Research and Training; Integrated Services Branch, Division of Services for Children with Special Health Needs.
2. Provide grants to States for professional training on genetic topics.
 - a. Give top priority to staff of State genetic and MCH programs.
 - b. Give priority to other primary care providers.
 - c. Give attention to cultural and ethnic diversity in the workforce.
 - d. Support development of core competencies for professionals.
3. Promote linkage of newborn screening to programs for CSHCN.
 - a. Use Federal guidelines and grants to encourage State-based efforts.
 - b. Provide technical assistance and incentives to State programs.
 - c. Require State-level tracking and performance monitoring.
 - d. Use the National Agenda for CSHCN and 2010 objectives in these activities.
4. Improve managed care practices.
 - a. Promote use of managed care contract specifications under development.
 - b. Fund demonstration projects between centers and managed care organizations.
5. Set consistent standards and guidelines among Federal agencies.
 - a. Promote consistency among HRSA, NIH, Health Care Financing Administration [HCFA], Agency for Health care Research and Quality [AHRQ, formerly Agency for Health Care Policy and Research], and Substance Abuse and Mental Health Services Agency [SAMHSA].
 - b. Encourage allocation of funds to be used for translating genetic science into practice.

Meeting One

6. Promote data collection, analysis, and reporting.
 - a. Support a process to define “core” data items.
 - b. Support research to study the impact on data systems of confidentiality, privacy, and health information protections.
 - c. Support research to improve understanding of families experience as consumers.

7. Protect treatment benefits in any new children’s health program.
 - a. Provide MCHB leadership to document the importance of comprehensive coverage.
 - b. Monitor the adequacy of benefits in SCHIP plans not using Medicaid benefits [MCHB and HRSA].

8. Provide support to advance genetic literacy for the public.
 - a. Focus on common, complex diseases and genetic variation.
 - b. Focus first on school-age populations and low-income families.

9. Develop genetic performance measures for Federal and State programs.
 - a. Use existing projects to define and refine measures/benchmarks.
 - b. Provide guidance to states and financing according to performance/competency.
 - c. Promote parent involvement in performance measurement.
 - d. Use the National Agenda for CSHCN and 2010 objectives in these activities.

10. Fund States to advance policy development, performance monitoring, and appropriate systems of care to serve children with genetic conditions and their families.

★ ★ ★

Partnerships with Consumers

Genetic Services Branch [GSB-MCHB-HRSA] emphasizes and promotes partnerships with families as consumers as part of its mission and objectives. Meeting One participants specifically recommended that GSB:

- Require inclusion of families in local, State and national meetings.
- Facilitate consumer involvement with special consideration for financial and physical barriers.
- Bring experienced consumer partners to monitor new partnerships.
- Provide technical assistance to states about how to collect information from families.
- Be respectful of privacy issues and concerns in data collection.
- Use integrated program approaches to reduce fragmentation of family services.
- Give families a forum to market their resources and capacities.
- Assist consumers in efforts to reach out to health professionals.
- Make sure all funded projects with websites are compliant with the Americans with Disabilities Act [ADA].
- Involve consumers in all training and education programs.

Priorities for HRSA Action

Meeting One participants recommended that the Genetic Services Branch-MCHB-HRSA use a proactive and forward-looking approach for supporting public health agencies, families, and health professionals. Participants encouraged the Genetic Services Branch to promote coordination and integration, use the Year 2010 framework and National Agenda for CSHCN, facilitate information sharing, foster public-private collaboration, encourage innovation, be “value added,” and be evidence based. The priorities of 26 individuals who provided written suggestions at the end of the meeting (in order of frequency mentioned) are shown below. These priorities apply to future activities of the Genetic Services Branch. Not surprisingly, the highest priority was given to Federal funding for State genetic and newborn screening program activities.

