

# **The Road Map to Implement Long-Term Follow-up and Treatment in Newborn Screening**

For the Subcommittee on Follow-up and Treatment of the Advisory Committee on Heritable Disorders and Genetic Diseases in Newborns and Children

## **A Meeting Summary**

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## **I. Background**

The Children's Health Act of 2000 authorized expanded research and services for a variety of childhood health problems. Title XXVI of the Children's Health Act of 2000, "Screening for Heritable Disorders," established a program to improve the ability of states to provide newborn and child screening for heritable disorders. This title enacted three sections of the Public Health Service (PHS) Act: sections 1109, 1110, and 1111. Under the last section, the Secretary of the U.S. Department of Health and Human Services (HHS) was directed to establish an Advisory Committee on Heritable Disorders in Newborns and Children. HHS expanded the Committee to include genetic diseases and renamed the Committee accordingly.

The Advisory Committee on Heritable Disorders and Genetic Diseases in Newborns and Children (ACHDGDNC) was established to assist the Secretary of HHS by providing (1) "advice and recommendations concerning the grants and projects" authorized under the Heritable Disorders Program and (2) "technical information to develop policies and priorities for this program that will enhance the ability of the State and local health agencies to provide for newborn and child screening, counseling and health care services for newborns and children having or at risk for heritable disorders."

Specifically, the Committee was charged with advising and guiding the Secretary on "the most appropriate application of universal newborn screening tests, technologies, policies, guidelines and programs for effectively reducing morbidity and mortality in newborns and children having or at risk for heritable disorders."

The ACHDGDNC began convening in 2004. As part of the continuing effort to carry out its charge, the National Newborn Screening and Genetics Resource Center, together with the Genetic Services Branch within the Maternal and Child Health Bureau of the Health Resources and Services Administration (HRSA), organized and hosted a gathering of experts in Bethesda, Maryland on April 18, 2007 to discuss developing and implementing a system for long-term follow-up care for children with health problems identified through newborn screening.

## II. Purpose of the Meeting

R. Rodney Howell, M.D., Professor of Pediatrics at the University of Miami and Chair of the ACHDGDNC, opened the meeting by pointing out that Long-term Follow-up (LTFU) is the biggest challenge in screening. Since most missed cases result from inappropriate follow-up and testing and mistaken diagnosis, only longitudinal studies can provide a solution. He introduced several of the themes that were elaborated during the day. He referred to specific programs in New England and in Region 4, the Southeast, that publish much information on laboratory performance to make sure all patients are reviewed, and to efforts in California on data collection and new computer systems, and he pointed to the importance of local and regional initiatives for the entire nation. Similarly, he introduced the theme of research efforts in regional collaborative networks and that of reimbursable activities — also topics that were to emerge in the day's deliberations.

Coleen Boyle, Ph.D., Director of Birth Defects and Developmental Disabilities, CDC/CCHP/NCBDDD, addressed the working group next, explaining that she is scheduled to report on the outcome of today's meeting to the full Advisory Committee, the ACHDGDNC, on May 17, 2007, along with the authors of the White Paper. The charge for today's working group, is to provide guidance on the White Paper, which was drafted between mid-February and mid-April of this year and is a preliminary attempt intended to take a bird's eye or even higher aerial view of the subject. She described the plan for the meeting: for the morning, she charged the group to use the draft White Paper as a framework to develop consensus on the definition, goals and components (or elements) of long-term follow-up of NBS. She indicated that once that was achieved, the remainder of the day would focus on the key participants in LTFU and begin dialogue on their major roles and responsibilities. She described the process by which the Working Group was created, pointing out that the working group members come from various parts of the country and bring to bear knowledge, background, and experience relevant to many aspects of the issue: they represent various professions (e.g., medical specialists in specific diseases, academic researchers, developmental professionals, pediatricians, clinical geneticists, health care attorneys) and many different settings (e.g. professional groups, health departments, foundations and centers, regional advisory committees, the Centers for Disease Control [CDC] and the Genetic Services Branch of HRSA) Individual introductions made clear the professional and personal involvement that members of the Workgroup have with these issues.

### **III. Introduction of the White Paper**

Alex R. Kemper, M.D., M.P. H., M.S., Director, Program on Pediatric Health Services Research, Department of Pediatrics and Duke Clinical Research Institute, Duke University, reviewed the White Paper on behalf of himself and his two co-authors: Stephen M. Downs, M.D., M.S., Director, Children's Health Services Research, Indiana University School of Medicine, and James Figge, M.D., M.B.A., Medical Director, Office of Health Insurance Programs, New York State Department of Health. Dr. Kemper said the draft represents a collaborative effort that involved considerable consultation but he emphasized the preliminary nature of the document and underscored that the ideas, comments, and criticisms to emerge during the day would be welcome.

#### ***Overview of Draft White Paper Definition and Goals***

Describing the authors' collaborative, incremental approach, Dr. Kemper outlined their specific objectives in preparing the White Paper:

- Define LTFU, including its goals and components (or elements).
- Develop a conceptual framework for LTFU (“model”).
- Describe important barriers to LTFU.
- List participants in LTFU as a first step towards future discussions of roles and responsibilities for all who would be involved.
- Deliver potential solutions. (He noted that technology is a tool, not a solution, and that it may be one of the easier elements to deal with.)

He pointed out that LTFU is not easy, and suggested that the overarching goal of LTFU is to achieve the best possible outcome for children and their families. (The Working Group agreed and at various times during the day indicated its wish to see this point featured prominently in the revised White Paper.)

He reviewed the initial draft of activities involved in LTFU and suggested that the process should extend from the moment of diagnostic confirmation to death. It was pointed out for the practical purpose of this meeting that the White Paper and this meeting was planned to consider LTFU only to age 18, and he recognized this is an arbitrary cutoff. (The working group later expressed some challenges with the categorical nature of this cutoff and wished to include some more nuanced observations about transitions during the entire lifespan.) He reviewed the Draft White Paper “Goals of LTFU” that were to be used as a starting point for discussion of the Workgroup in development of consensus on goals and components (*see later in meeting summary for consensus*).

#### ***Draft White Paper Goals of LTFU***

The draft goals are three-fold:

- (1) Access to care
- (2) Care coordination (including quality improvement)
- (3) Expanding the evidence base

### *Models for LTFU*

In reviewing some models for LTFU, Dr. Kemper mentioned the following:

- The medical home: a partnership approach with families to provide health care that is accessible, family-centered, coordinated, comprehensive, continuous, compassionate, and culturally effective.
- The Children's Oncology Group (COG), which provides a model for some but not all of the LTFU process. He pointed out that COG model does not address treatment and therefore is not perfectly applicable to this effort.
- The Chronic Care Model, a good one for looking at various conditions and one that illustrates the relationship between the community and the health care system writ large and how they can work together to develop a supportive, integrated community. The authors considered this a fine general model but not one for newborn screening, insofar as it offered no clear role for public health, which is subsumed into the health care system. Dr. Kemper pointed out that for newborn screening, the public health component is very important (a view shared by the working group, as later discussions would demonstrate).

He offered the specific model in the Draft White Paper as a framework for discussion.

### *Draft White Paper Barriers to LTFU*

Dr. Kemper reviewed some of the barriers to LTFU, and suggested that some barriers focus on the individual, others on the condition, and others on the health care system.

Some barriers involving **the individual** are: (1) protection of privacy and human subjects, (2) transitioning through adulthood, (3) potential risks to insurability, and (4) timely dissemination of information.

There are barriers involving **the condition**, for example: (1) the heterogeneity of conditions and (2) whether the condition is phenotypic (determined by the description of the physical and behavioral characteristics of the organism) or genotypic (determined by the description of the actual physical material made up of DNA passed to the child by its parents at the child's conception).

There are barriers related to **the health care system**:

- Varying infrastructure
- Variations in workforce capacity

- Lack of workforce preparedness
- Lack of consistency from state to state
- Variation in users
- Lack of standards
- Lack of electronic health record (EHR) interoperability
- Role of pharmaceutical/device manufacturers (raising many proprietary issues)
- Differences in interest among stakeholders (e.g., individuals, health departments, companies)

### ***White Paper Draft List of LTFU Participants***

Dr. Kemper reviewed an initial list of the key participants including public and private entities: Coalition For Informed Choice (CFIC), primary care providers, specialty care providers, local public health agencies, state public health agencies, private insurers, Medicare, Medicaid, the State Children’s Health Insurance Program (SCHIP), Regional Genetic and Newborn Screening Services Collaboratives, National Coordinating Center, National Newborn Screening and Genetics Resource Center, HRSA, the Agency for Healthcare Research and Quality (AHRQ), the CDC, the National Institutes of Health (NIH), advocacy groups and other nonprofit organizations.

### ***Initial Plenary Discussion of White Paper***

Drs. Kemper and Boyle opened the discussion of the Draft White Paper. Dr. Kemper urged the group not to be discouraged by the barriers, and he and Dr. Boyle invited attendees to begin the process to develop consensus on goals and components (or elements) of LTFU and also to begin to consider potential models. The discussion began with a plenary discussion, and Dr. Boyle described plans for the breakout sessions, and for the afternoon session in which the workgroup would begin to consider the list of participants.

The full working group’s initial responses to the broad array of topics in the White Paper served to introduce some of the themes that the smaller breakout groups would explore in greater depth later in the day.

Two participants addressed the notion of *transitions*. While acknowledging that individuals over the age of 18 (or possible 21) fall outside the purview of this meeting (based on the charge of the Advisory Committee to which this WG reports), they felt it important to find a way to include the full life span in the logic model.

Others mentioned *insurability* and *financial implications* as issues that need attention. In this connection, one participant suggested that it might be helpful for the working group to focus first on what needed to be done and to address the cost implications at a later time.

Six other points emerged from this discussion. The White Paper should: (1) be clear that LTFU includes treatment; (2) be clear that LTFU includes expanding the evidence basis; (3) avoid the use of terms that lack definition or have competing definition; (4) include the important role of

the public sector and the essential notion of a public-private partnership; (5) explore more fully some existing models (such as COG and Cystic Fibrosis [CF]), especially in connection with the involvement of public health; and (6) remember to consider legislation and the important factor of public policy in moving the LTFU agenda forward.

## IV. Small Group Breakout and Summary Plenary Discussions

The full working group broke into three smaller groups, each meeting for two and a quarter hours. Minutes of the nearly seven hours of conversation contained some specific page references and suggestions for individual word changes and were forwarded to the authors of the White Paper. What follows is a summary of the group morning conversations, afternoon presentations to the full assembly, and plenary discussions of key concepts and questions, organized thematically.

### *Definition and Goals of LTFU*

The attendees came to consensus on the basic definition and goals of LTFU: To achieve the best possible outcome for children and their families.

In addition, the WG suggested:

- Begin the White Paper with a clear statement of values.
- Define terms (such as “medical home”) clearly. In the context of medical home, include the idea that there will be an intentional conversation about who is taking responsibility and explore the various different relationships among the care providers, the family and the community.
- Be cautious in use of terms that have variable meaning. In particular there was concern about the meaning of the term “care coordination” and need to define care coordination activities separately from those of clinical treatment.
- Define outcome measures including for those with proven disorders and those with suspected disorders or false positive screens, and consider criteria for who should be followed (e.g. biochemical diagnosis vs. phenotypic diagnosis). In particular, outcome measures should include:
  1. Quality evaluation/surveillance (include tracking to make sure the patients receive treatment)
  2. Clinical and public health research including the collection of observational data on the outcomes of therapy.
- The WG emphasized that clinical care/treatment (including treatment specific to the disorder and preventive care, health promotion, disease prevention) is a crucial element of LTFU.
- Care coordination including development of a written management plan was identified also as a critical element.
- Scope: focus on age 0 to 18 but include the whole life span. LTFU must include the major milestones (puberty, childbearing, and the like). This concept of *transitions* is important in LTFU. Health and functional outcomes include transitioning to the adult health care system. Furthermore, making sure that a person can achieve life goals and

individual potential extends beyond age 18. Current legislative mandates put age limits on formula/foods, setting a poor precedent. LTFU should take a developmental lifespan perspective.

### ***The White Paper Model and its Relation to Other Models***

- The paper should put more emphasis on clinical care. More work is needed on translating findings into what they mean for treatment. The LTFU model should make clear that improvement in care is integral to LTFU.
- Consider to what extent LTFU for newborn screening is different from that for other children with special health care needs. For example, there are genetic ramifications for the family raising the issue of voluntary vs. mandatory screening.
- LTFU should put the patient, and not the health outcomes, at the center.
- The LTFU model should be generic, even though there is a need for evidence based on specific diseases. A primary care physician (PCP) would need a single, generic model but it should not supplant disease-specific guidelines, where they exist.
- The LTFU model must include the public health aspect: surveillance, data collection, and integration with other databases from the public health sector. And in this context, the model should emphasize the notion of *partnership* between individual caregivers and public health elements. This is not an issue unique to newborn screening (it arises with all chronic diseases) but this LTFU project is an opportunity for the subcommittee to emphasize the partnership between the care delivery system and the public health system and to develop a model for an effective partnership.
- Acknowledge other models and do not imply that this is an entirely new idea. Make a greater effort to learn from the COG and CF models.
- The LTFU model is a *hybrid* model, combining the chronic care model and the public health function.
- Because there is a need for a mechanism to disseminate best practices, the feedback loop — which would be a means of self-improvement — should be built into the model.
- In terms of Continuous Quality Improvement (CQI), explore what the Institute for Healthcare Improvement and the National Initiative for Healthcare Quality have done.
- Generally speaking, include ethical, legal, and social issues.

### ***Treatment and Care Coordination***

- Consider terminology. (Also see previous summary of related discussion.) Attendees discussed whether treatment should be considered as part of care coordination or should the category be called care and care coordination, or perhaps care and service coordination? Terminology aside, the groups agreed that LTFU should include treatment. Some attendees suggested that care/treatment and care coordination are not discrete notions, and that the quality of care coordination may determine the quality of care: it could be, for example, that children in a state with the best care coordination fare the best.

- Models and systems must acknowledge and address problems with access to some services. For example, children with some disorders are not considered “disabled” for the purposes of Medicaid and therefore do not have access to early intervention and a social worker who tracks them.
- The family needs a single point of contact. The LTFU model must emphasize a comprehensive focus for the clinical/medical home, which needs to include developmental, medical, mental health, and education/support elements.
- Some discussants envisioned a personal health record, Web-based and interoperable.
  1. This raised many issues, primarily: who will maintain it and who will have access to it? A current project of the Research Triangle Institute (with which Dr. Figge is familiar) is addressing many of these issues.
  2. The matter of data standards is important. There is a need for a shared vocabulary and a mechanism for exchanging the data. It is very difficult to capture the data at the point of care. It is quite easy to capture laboratory data and relatively easy to capture pharmaceutical data. Diagnostic data need a common designation. This LTFU project is an opportunity to work on data standards and the issue of who gets to see what and when. The data elements must be compatible with other systems. (In this connection, Dr. Howell pointed out that the Secretary of HHS is very interested in electronic health records and is convening a meeting on the subject.)
  3. Collection of data is important for CQI.
- In the context of LTFU, perhaps the United Kingdom’s “virtual centers” could offer examples of how to proceed. The UK has designated geographic areas over which a physician is responsible even if the physician does not deliver all of the care.
- Should there be regional coordinating centers? Disease specific? It was observed that the coordinating center should be developed on the basis of the uses to which it is going to be put. For a very rare disorder, a national database might be preferable. For Quality Improvement, a regional or local database may be better. (Working group members frequently expressed hesitation about national databases, which can raise objections from many quarters. Some said that a national approach might be avoided if there is agreement on data standards and elements and on access.)
- The point was made that the LTFU model for treatment and continued improvement of treatment and care coordination should try to find a way to include “hunches” formed on the basis of anecdotal evidence, an approach that has been driven out of our current health care system. There needs to be a way to take advantage of the long-term experience of practitioners by being attentive to those in the field, capturing anecdotes, and trying to identify patterns.
- Effective treatment and care coordination depends on the extent to which the caregiver has access to cutting edge developments.
- We may need a federal policy on establishing access to medical centers which lie outside the patient’s state or outside the private insurance network of providers.
- Some other specific suggested strategies for treatment and care coordination were:
  1. The plans should be written.
  2. Develop templates for diseases; these can then be personalized.
  3. Train and hire people to do care coordination. The system should not rely on the PCP, the specialist, or a registered nurse. The task needs a fulltime person.

4. Care coordination should be across systems and services and include community, the medical system, and the public and private sectors.
5. Intervention should include school systems.
6. Data collection needs to track individuals as they move geographically and through systems.
7. Treatment and care coordination should encompass the entire lifespan.

### ***Role of the Family***

- LTFU must include active family involvement. The *perceptions* of family members can be important. Two centers with the same protocol may differ in outcomes based on the personal interaction between provider and patient/family. *The experience* of family members can be important. Family members can become the real experts in the medical condition.
- There is a need for provider training on how to partner with families, which — like so many other suggestions — has financial implications.
- Family support groups and patient advocacy groups can play a bigger role in the partnership. (Parent groups were instrumental in the progress made in newborn screening.)
- Family involvement raises such matters as the ability to pay and the constellation of issues around genetics.
- Some problems related to the family are: (1) language and culture, (2) family location and mobility, (3) family priorities connected to such concerns as employment and transportation, and (4) the family's relationship to the PCP.

### ***Role of the Public Health Sector***

- Mandated screening ties LTFU into the public health system (the state public health agencies and the HRSA Title V agencies). This makes it different from the chronic care model.
- As included under the “Treatment and Care Coordination” rubric above, the important evaluation/surveillance component of LTFU is a public health function, because of the need to address related funding, privacy, and genetic discrimination issues. It is therefore perhaps in the area of evaluation that public health can play its greatest role.
- All newborn screening is under the jurisdiction of the state. LTFU efforts should have a national component. While regional efforts could be a model for framing national legislation, realism dictates an awareness that sweeping changes to our country's health care system are unlikely. Therefore, perhaps the goal should be to enact similar state-level legislation throughout the country, mandating that some reporting of data is fair game and addressing privacy issues. (The federal government offers examples of how to deal with privacy issues.)

## *Manpower Issues*

- Access to an adequate, capable workforce is essential to providing good clinical care. The term “access” is frequently used in the context of needed financial resources. In the context of LTFU, it may mean getting to needed expertise.
- More skilled providers are needed. There are not enough medical specialists and other health care providers and they are poorly distributed nationally. In some cases there may be only one expert in the country.
- Genetic specialists and counselors play an important role, and more are needed.
- The subspecialist cannot function as the PCP and the PCP cannot function as the prime care coordinator.

## *Research*

- LTFU must contain the idea that care improvement is an integral part of the process, and much of care improvement depends on research.
- There are various aspects to research: (1) laboratory to bench to syringe, (2) syringe to community, and (3) structural research to determine which components work best in the delivery of health care.
- Research includes: (1) clinical trials with new therapies and (2) collection of observational data. The two are complementary and provide a platform on which to build new knowledge.
- If families perceive clinical studies as part of treatment and not just research, they will opt in. (Dr. Howell gave the example of experience with Pompe Disease.)
- There is a need for evidence on which to develop clinical guidelines. Because many of the disorders are rare, we need a national protocol to develop the guidelines. There is a need to study long-term outcomes of various treatments.
- We need to improve the evidence and disseminate best practices. Currently only a few people know the evidence; we need a mechanism for dissemination.

## *Financing*

- Financing cuts across almost all issues. A new model is needed to support the comprehensive, interdisciplinary team approach and care coordination, with state- and federal-level solutions. The group’s documents must make clear the financial implications of the needs of LTFU.
- The White Paper may address the issues of financing models. (An example of one restriction is the refusal of Medicaid and some insurance companies to pay for a patient to see more than one health care provider on a single visit.) If the best approach to LTFU is multidisciplinary, how can the team approach be financed?

- Our current system does not reimburse a function that may be vitally important to the family and that represents a single contact who provides answers to a variety of straightforward questions such as: “May I have a new prescription?” and “My child is vomiting; what should I do?” Some states have solutions to this and others do not. LTFU needs to emphasize the need nationally.
- The current system does not pay for self-management training.
- The current system pays for performing procedures, not for such vital aspects of care as explaining, discussing, hand-holding.
- At present, there is a skewed relationship between institutions and other parts of the system. The cost of providing services is borne by institutions but if there is a dire outcome, those costs may be incurred by someone else altogether. Incentives are crossed: the great financial pressures on institutions make it difficult for them to take a broader perspective and consider economies to the system as a whole rather than to the institution. As things stand now, the cost of most LTFU falls to referral institutions, primarily universities. Even the diagnostic confirmation is borne by these institutions. They have a moral responsibility to treat individuals but no way of recovering costs.
- LTFU needs a coordinated team approach. The smaller the team component, the less able it is to carry the financial burden of the piece of the coordinated system which it has been assigned if it cannot support that service with its own budget.
- The group might consider developing model legislation at the state level.

## **V. Plenary Discussion**

After the presentation of small group reports, Alan R. Hinman, M.D., M.P.H, Senior Public Health Scientist, Task Force for Child Survival and Development, Public Health Informatics Institute, facilitated a general discussion. Dr. Hinman clarified the objective of the discussion, which was to develop, beginning with the Draft White Paper list, the participants in LTFU and to begin to explore the roles and responsibilities of various players with respect to the components of LTFU, recognizing that — in the time available — these listings could not be exhaustive. The group then worked systematically to begin to describe these roles and responsibilities, as follows:

### ***Affected Individuals: children and their families with identified conditions (CFIC)***

- CFIC should participate from the start in planning and setting goals, sharing in the decision making as a partner although not, of course, as a professional care provider.
- LTFU should include support from other affected families. This could come from an outside support group or through a one-on-one relationship. It might include advocacy groups and other nonprofit organizations. Online support groups are active and effective.
- A good model to follow is the patient's rights and responsibilities posting in every hospital.
- CFIC have a responsibility to comply with recommendations. Much education and communication has to precede this. Both sides need to demonstrate a willingness to learn.
- CFIC have a role in advocacy. Can this be called a responsibility? In reality, they are the only players who can really affect legislation.
- They have a responsibility to keep their information current.
- They have a responsibility to provide feedback.

### ***Primary Care Providers (PCPs)***

The PCP should:

- Become familiar with the disorders of their patients, learning about the social as well as medical impact of the illness and situation.
- Be willing to understand and acknowledge limitations.
- Offer (although not necessarily provide) a medical home; there should be explicit discussion with CFIC on this subject.
- Operate within the standards of care.
- Listen to CFIC and be responsive to their needs.
- Assure that families have up-to-date information about the condition.
- Be ready to provide referrals, as appropriate, establishing a partnership with a subspecialist in a two-way relationship, and demonstrate a willingness to co-manage.

- Advocate for the patient, communicating with insurance companies and other financing entities in a timely way.
- Be willing to contribute to the knowledge base.
- Sometimes have a role in enrolling patients in clinical trials.
- Recognize their role in the newborn screening program, responding to queries from it and confirming that the family has completed appropriate genetic counseling. This is part of a coordinated written plan (see next item).
- Establish and update a written care plan. (An Individualized Family Service Plan [IFSP] documents and guides the early intervention process for children with disabilities and their families. It is the vehicle through which effective early intervention is implemented in accordance with Part C of the Individuals with Disabilities Education Act [IDEA].)
- Explicitly identify the locus of care coordination — in partnership with the family, the specialist, and the state health department — and participate in the care coordination.
- Collaborate with the local community hospital.
- Coordinate the transfer of care when the patient moves and when the patient transitions.
- Be familiar with emergency care plans, which differ from written care plans.
- Assist in arranging for local delivery of needed medications and/or care.
- Respond to CFIC, to specialists, and to public health officials (by, for example, participating in surveys).

Dr. Hinman underscored that some of the items listed above can be repeated under the rubric of roles and responsibilities for specialists as well as for care coordinators. Some group members asked how many of the above-listed activities a PCP with a busy practice could take on. They acknowledged that where there are no subspecialists, the PCP might need to take on more. Most of the above-listed items describe the elements of providing a good medical home.

### ***Specialty and Subspecialty Providers***

The items listed below are in addition to the ones listed under the primary care providers. In compiling them the group introduced the concept of “consultant,” pointing out that there are different levels of consulting. Sometimes the consultant’s contributions are ignored, but that is by no means always the case.

The specialist or subspecialist should:

- From the start, participate as a partner in overall care.
- Acknowledge the important substantive role played by the PCP and the family.
- Communicate and respond to the PCP in a courteous and timely way.
- Recognize the limitations of the PCP in terms of time and resources and provide support to the PCP.
- Provide leadership for developing a care plan for the specific condition. At a state level, the specialist or subspecialist should keep PCPs informed. (It was noted that in California, for example, they have played a leading role in developing public policy.)

- Continue to ensure that the patient has a medical home (for example, finding a PCP where there is none).
- Keep abreast of the current state of knowledge.
- Assure that appropriate genetic services, especially counseling, are provided.
- Acknowledge and accept a pivotal role in education.
- Be willing to participate in regional and national data collection and in clinical trials.
- Be willing to serve as a consultant to other care providers.

### ***Public Health Agencies***

Members of the working group commented that there is a widespread lack of consensus about the role of public health agencies in supporting children and families faced with these medical situations.

Public health agencies should:

- Provide reliable and timely newborn screening.
- Understand outcomes in order to influence services.
- Accept responsibility to assure short- and long-term follow-up.
- Assure information systems that are adequate for supporting LTFU and aggregating data for inter-state comparison.
- Consider what goes to short-term follow-up and what goes to long-term follow-up and assure internal advocacy for necessary LTFU.
- Play a role in the quality improvement of LTFU.
- Disseminate information.
- Stay up-to-date on developments in screening and provide ongoing education to the public, to professionals, and to policy makers.
- Educate the public about the importance of LTFU.
- Participate in developing national standards, definitions, and data elements necessary for LTFU.
- Participate in care coordination and, in some settings, provide the coordination. (Responsibility for coordination may vary from place to place. Multiple models exist.)
- Support the PCP, specialists and subspecialists, and care coordinators by providing, among other things, technical assistance and resource information.
- Partner with other programs and professionals to ensure that LTFU is appropriate and adequate.
- Communicate with families to determine how their care is progressing. (This can contribute to CQI.)
- Analyze and interpret data; evaluate and monitor LTFU and, again, assure the dissemination of information.
- Coordinate information on patient and family decisions about participation.
- Educate health plans and health insurers and advocate for appropriate coverage.
- Assure the free flow of information among authorized health care providers.
- Recognize the regulatory force of public health agencies.

Dr. Hinman, forced by the hour to conclude the discussion, commended the working group for its excellent work in beginning to list the participants and begin to consider the roles and responsibilities of the various players in LTFU. He reviewed several suggestions that emerged during the deliberations: (1) explore a federal mandate for surveillance and tracking, (2) consider developing a model for state or federal legislation to support the care and delivery infrastructure, (3) affirm that there should be some federal standards for personal health records, (4) define a standard of care that could be a means of obtaining resources, and (5) include genetics in Title V of the Social Security Act, administered by HRSA. (Title V has been amended many times over the years to reflect the expansion of the national interest in maternal and child health.)

## **VI. Next Steps and Wrap-Up**

Drs. Boyle, Kemper, and Downs all concurred that LTFU is very challenging. Drs. Kemper, Downs, and Figge will now use the observations and suggestions from members of the working group to revise the White Paper. They will circulate the revised draft to the subcommittee and to working group members to get their reaction. All this is to be done before May 17, 2007, the day when Dr. Boyle and the White Paper authors are to report to the full Advisory Committee, the ACHDGDNC.

In conclusion, workgroup participants *reiterated* that the White Paper should set a tone from the start by communicating how the group feels about children and families. Newborn screening is not just a test. It is a broad enterprise, of which LTFU is an essential part, and an enterprise that involves many players who need to work in partnership. Collaboration is the name of the game.

## **Appendix: List of Participants**

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# The Road Map to Implement Long-Term Follow-up and Treatment in Newborn Screening

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Bethesda, MD  
April 18, 2007

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