

**Project Title. The New England Regional Genetics and Newborn Screening Collaborative (NERC)**

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Problem: Newborn screening in Region 1 has expanded recently in response to increased technical capability of tandem mass spectrometry and to national leadership advocating for expansion. This project will build upon our experience with regionalized newborn screening to enhance genetic service capacity for genetic conditions identified by newborn screening and those identified clinically in Region 1. The Mission of the New England Regional Genetics and Newborn Screening Collaborative (NERC) is to promote and improve health and social well-being of those with inherited conditions through collaborations among public health professionals, private health professionals, educators, consumers and advocates.

The NERC will address the following goals:

- Goal 1: Establish and maintain a regional center for genetics and newborn screening.
- Goal 2: Collaborate to facilitate access to genetics services, expertise and technology particularly for underserved populations and in rural areas.
- Goal 3. Develop effective practice models for adolescents and young adults with genetic conditions who are transitioning from pediatric to adult health care.
- Goal 4. Develop effective partnerships to further Medical Home practices in the region.
- Goal 5. Serve as the focal point for effective genetics education and dissemination of genetics information.
- Goal 6. Demonstrate effective collaborations with other regional and national stakeholders.
- Goal 7. Examine the relevant public policy and ethical, legal and social issues affecting individuals with genetic conditions, their families, and health care providers and educators.
- Goal 8. Utilize a small project program to accomplish unanticipated and innovative activities that emerge within the region.
- Goal 9. Provide opportunities to increase discussions with school systems and state special education leaders to improve access and support to educational services in the least restrictive environment.
- Goal 10. Complete both quantitative and qualitative evaluations of processes and outcomes of all goals, activities and projects undertaken by the NERC.

Methodology: The NERC will employ the logic model to address each of the goals. The work will be accomplished by Working Groups, a Collaborative Council of Working Group Leaders, the Regional Coordinating Center, and an Advisory Committee. An independent evaluator will conduct both formative and summative evaluations.

## I. INTRODUCTION

This application is in response to HRSA-07-016 Program Guidance, Project 2: “Regional Genetic and Newborn Screening Service Collaboratives” for Region 1. The purpose of the New England Regional Genetics and Newborn Screening Collaborative is to improve the health of those living with genetic disorders in Region 1 and nationally. This will be accomplished through specific and formal collaborations among: public health officials, consumers and advocates, primary health care providers, specialty health care providers, and, other agencies and individuals who serve as formal and informal supports to those with genetic conditions.

The primary goal of The New England Regional Collaborative (NERC) is to assure that individuals with genetic disorders and their families have access to quality care and appropriate genetic expertise and information in the context of a medical home that provides accessible, family-centered, continuous, comprehensive, coordinated, compassionate, and culturally effective care.

The new structure of NERC as proposed is specifically designed to strengthen communication and collaboration among public health professionals, individuals, families, primary care providers, and genetic medicine and other subspecialty providers. As NERRG, Inc will no longer serve as the Regional Coordinating Center, the Regional Coordinating Center (RCC) at the Institute on Disability, University of New Hampshire (UNH) has been established to serve as “headquarters” for the NERC. The RCC also will quantitatively and qualitatively evaluate outcomes of projects undertaken to accomplish the stated goals. The NERC will establish and maintain an Advisory Committee that will provide overall direction for all project activities. NERC will have eight Working Groups to address specific goals and objectives. The leader of each working group will be a member of the Collaborative Council which will address collectively any barriers that might arise in our collaborations. The Project Director and the RCC will lead, coordinate, administer and evaluate all aspects of the NERC. The Project Director, Co-Director and Project Coordinator will communicate with the HRSA/MCH/GSB Project Officers appropriately to assure that all aspects of this Cooperative Agreement are addressed. Also, the Project Director and Project Coordinator will work with the National Coordinating Center, other Regional Collaboratives and all key stakeholders.

The proposed goals and objectives of the NERC are the result of work by the current cooperative agreement (New England Regional Genetics and Newborn Screening Collaborative HRSA Grant # 1U22MC03959), the emerging New England state plans, seven recent teleconferences of the NERC Planning Committee and two meetings of the NE Regional Genetics and NBS Planning Committee at the 2006 annual meeting of NERGG, Inc. This Planning Committee work has incorporated successes made to date during the last three years. The NERC Planning Group has committed to a 2-day planning retreat in Spring 2007 to finalize the NERC Regional Plan for our June 1, 2007 anticipated start date for this cooperative agreement.

The Northeast Region I, consisting of the six New England states is varied by race and ethnicity, socioeconomic status, rural and urban living. There are approximately 173,000 births each year and population of 13,922,517 in New England. Diversity in demographics and health care options results in variable distribution of services across the region.

Nevertheless, there have been notable regional genetic activities. The first of these activities was the 1977 regionalization of newborn screening (NBS) with the establishment of the New England Newborn Screening Program (NENSP). NENSP is one of the most comprehensive and effective NBS programs in the U.S. The NENSP, a regional service-based collaborative with a long history of coordinated services, regional grants and publications, is an excellent example. This program enters virtually all newborn infants into a system of NBS that covers 5 of the 6 New England states (the sixth state, Connecticut, conducts its own state screening program). The NENSP provides a variety of screening services (education, laboratory testing, result reporting, clinical consultation, outcome tracking and research) for all newborns in the five states.

The New England Regional Genetics Group (now NERGG, Inc.) was established in 1981 to bring together those consumers, public health professionals and clinical genetics professionals in the region to address common problems of access to quality genetics health care services and information. The New England Consortium of Metabolic Programs (NECMP), the New England Pediatric Sickle Cell Consortium and the Massachusetts Cystic Fibrosis (CF) Workgroup were formed to exchange information, organize NBS follow-up services, and conduct research on a regional basis. The New England Consortium of Metabolic Programs (NECMP) is comprised of representatives from each of the states in our region, including the directors of the 11 metabolic programs in New England and their associated staff, public health and newborn screening personnel, industry leaders, and several consumers. The purpose of the New England Consortium is to increase knowledge of metabolic disorders and to improve the delivery of health care to patients in our region.

The opportunity now is to build on these successes and to address barriers to effective collaboration. The New England Regional Genetics and Newborn Screening Collaborative will build on the NENSP, NECMP structures to create both state-based teams and to develop regional technical assistance capacity to support these state teams. The new Regional Coordinating Center at UNH will serve to assure that that state teams have access to all the resources of the region in developing solutions to local needs. The state teams, consisting of the NBS coordinator, genetics specialty provider, consumers/advocates, Federally qualified health center leaders, and medical home providers, will assure access to quality care and appropriate genetic expertise and information in the context of a medical home that provides accessible, family-centered, continuous, comprehensive, coordinated, compassionate, and culturally effective care in their states.

## **II. NEEDS ASSESSMENT**

Each New England state has conducted genetics health and needs assessment and planning although each state differs in its' progress toward formulation of a state genetics plan. Our goal is to establish a "shared baseline" for states as part of developing a regional plan for the NERC. The Regional Coordinating Center at UNH will facilitate the establishment of a shared baseline for the states in the region and to finalize and implement a Regional Genetics Plan. Each state has specific particularities that need to be addressed. The regional plan will establish the shared baseline to which each state may add plans to meet local, specific needs. The RCC will complete the regional plan by the start date of this cooperative agreement on June 1, 2007.

There are thematic areas of need that are common to the state plans. They include:

1. Public Policy and Ethical, Legal and Social Issues;
2. Practice Models and Delivery Systems;
3. Education, Technical Assistance, Dissemination;
4. Genetics Services; and,
5. Public Health Infrastructure.

#### **IIa. State Genetics Needs Summaries:**

*New Hampshire* convened a planning group in December 2006 to address specific needs. Present for the planning meeting were Judy Bumbalo, PhD, Title V Bureau Chief (NH Special Medical Services); Kathy Cahill, RN (Transition Group); Ruth Fox, RN, Coordinator, NH EHDI; Audrey Knight, RN, MCH; Marcia Lavochkin, Newborn Screening Coordinator; Debra Nelson, MED, consumer, NERGG, Inc Board Member and Institute on Disability/UNH; Terry Olson-Martin, NH Family Voices; and Ellen Wheatley, Child Development Bureau, NH DHHS. The NH planning group identified the following needs to be addressed by the RCC:

1. Provide TA and support to develop a regional genetics plan that NH and other states can adopt/adapt;
2. Provide opportunities for networking among states, both discipline-specific and cross-discipline;
3. Facilitate a meeting of all partners on collaboration (what does it mean? how can we best collaborate?).

Regarding NH practice models and delivery systems, the planning group identified the following activities:

1. Further develop the NH model for youth health care transition (NH Youth Health Care Transition Project) and the Youth Health Care Transition Coalition;
2. Provide mini-grants to family organizations (Family Voices) for family education;
3. Explore the applicability of the healthcare consultant model (DHHS provides TA/consultation from nurses to early care and ed. programs; including childcare) and health manager model used by Head Start;
4. Promote the adoption of the Medical Home model by adult providers caring for youth/adults with chronic health conditions/disabilities;
5. Provide support to states for health care transition coordinators to build capacity in medical practices to care for youth/adults with chronic health conditions/disabilities.

Regarding the Ethical/Legal/Social issues:

1. Provide TA from ethical experts to address issues such as dissemination, equal access to specialty care, insurance discrimination;
2. Provide TA from legal experts regarding issues such as access to community programs (childcare, Boy/Girl Scouts, Head Start, schools, etc.);
3. Promote health literacy for families (including youth) and practitioners (e.g., how to talk with families and youth).

Regarding Dissemination

1. Support the Education Collaborative to continue its work;
2. Identify or develop fact sheets and/or packets for adult providers, pediatric providers and families on various genetic conditions (including implications for care); assuring that

materials are available in multiple languages and sensitive to diversity; NH needs materials for families on sickle cell/hemoglobin;

3. Promote networking among states to share information, materials and strategies;
4. Create (or identify) a genetic condition web page/site;
5. Survey adult providers regarding their needs in caring for young adults/adults with health conditions/disabilities;
6. Support the development of a jointly planned (i.e., by regional stakeholders) health care transition conference and offered in various locations accessible to providers and families in all states.

**Rhode Island** completed its last formal state Genetics Plan in 2002; recently, an internal survey for needs assessment has been completed:

Rhode Island Genetic Survey Conclusions [Only Mission one had questions addressing Assessment, Policy and Assurance. Mission Two only had questions addressing Assessment, and Mission Three had questions addressing Assessment and Policy only.]

Mission 1: Help people understand the genetic contribution to health

Assessment:

1. Better genetics awareness within HEALTH is necessary. The genetic resources in Rhode Island are not well known among the programs in HEALTH. A brief in-service or printed brochure of the resources and clinical services available in Rhode Island may be a way to educate the programs within HEALTH, thus allowing the basic knowledge to be passed on, as appropriate within the operations of the programs.

Policy:

1. Involve more consumers in all divisional activities that relate to genetics/genomics. Develop mechanisms for better, wider communication with consumer representatives. The number of parent consultants should be expanded to include programs beyond Family Health, especially within The Division of Disease Prevention and Control. Standardized resources for the parent consultants to use in their contact with the general public would be useful. These may exist for newborn screening, but may need to be developed for the adult conditions. Furthermore, these resources should be shared with any of the advisory boards where consumers are present so that the knowledge can be passed on to larger groups at a time.

Assurance:

1. The incorporation of genetics into any grants possible must be considered in all programs. As of now, there are a very small percentage of programs doing this. For the goals to be achieved in the state plan, funding must be available for genetic related projects. Also, programs should continue to consider more about how genetics can be incorporated into all aspects of outreach regarding: screening and prevention, diagnostic and therapeutic, and counseling, social services and family support. The Division of Family Health is the leader in incorporating genetics into their programs, but other Divisions, especially Disease Prevention and Control must join them in this effort.
2. The incorporation of family history into all applicable programs (within Family Health and Disease Prevention and Control) will be an important part of the future development of a genetics component into programs. The promotional materials, especially in Chronic Disease, need to include family history information.
3. Adult genetics needs to be brought up to the same level as newborn and children. Nearly all of the tasks that address services are for newborns and children, but the Division of

Disease Prevention and Control houses asthma, arthritis, obesity, tobacco control, cancer programs, diabetes, and women's health, all of which have genetic components.

Rhode Island Mission 2: Provide a forum to facilitate discussion of the impact of genetic knowledge and technology on communities and individuals and public health.

Assessment:

1. Evaluation of genetic services should be maintained and/or expanded within existing programs of all divisions, including Family Health.
2. There are many genetic conditions that affect adults and the potential barriers to accessing genetic services should be identified. The Division of Disease Prevention and Control should collect more info on family history and composition.

Rhode Island Mission 3: Apply knowledge and technology wisely to prevent disease and to protect and promote health.

Assessment:

1. The majority of programs indicated that they need additional support staff or expertise to achieve their goals and to include genetics in their programs. Again, including genetics in all grants possible will help to achieve this, especially the Division of Disease Prevention and Control for adult onset conditions.
2. The adequacy of private sector genetic resources was considered to have moderate to significant unmet needs among the Division of Family Health as well as a quarter of total programs within HEALTH.
3. In addition, the evaluation of genetic services should be expanded in the future where possible. Over one fourth of the programs in Family Health have plans for this.

Policy:

1. The HEALTH library is underutilized by programs within HEALTH and may be an indication that this resource is of limited use. Furthermore, the continued development and maintenance of the genetics website within HEALTH may be a more valuable link to refer consumers, providers, third party payers, academics, students and others.
2. Programs should be better educated about how they fit into the larger picture with regard to genetics and public health. It is clear that general genetics training is desired by the majority of programs, and this awareness of need should be recognized quickly. While genetic consulting staff may be able to provide much of the training, there is an additional source that may be of great use to HEALTH. An online program created by the Michigan Center for Genomics and Public Health entitled "Six Weeks to Genomic Awareness" is now available through the Centers for Disease Control and Prevention website. This online series of presentations is designed to provide public health professionals a foundation for understanding how genomics advances are relevant to public health. The series includes introductory genomics concepts, population genomics, genetic testing, ethical, legal and social issues, and lists state and national resources for public health professionals. In addition, a 45-minute introductory program, "Genomics for Public Health Practitioners" is also available for public health practitioners who have minimal experience in the area of genomics as it pertains to public health. This source may be the most efficient and swift way to encourage an introduction of genetics to personnel to be later followed up with more specific training and education.
3. A desire for further genetics knowledge and was particularly noted among the Divisions of Disease Prevention and Control, Family Health and Health Laboratories, where the higher percentages of need were found.

4. Once internal genetic education is addressed, the general public genetic awareness must be addressed. Almost one quarter of programs have already included genetics information as part of their community education efforts. In addition, approximately half of the programs further indicated that genetics could be easily integrated into their current community education activities. All divisions should explore more ways to integrate genetics.

Future Challenges:

1. Creating smoother transitions (EI to school systems, hospital to home, home to school etc.) among the 3-21 year old age group. The Division of Family health will be a key player in this area. The Health Services Regulation Division may also lend resources regarding hospital to home transitions as their area of expertise.
2. Environmental Health needs to develop a genetics component to their program as the gene/environment interaction is a key element to the development of disease.

The *Massachusetts* Department of Public Health (MDPH) has identified the following areas of focus for statewide public health genetics initiatives (2003-2005):

Professional Education and Training

1. Promote genetics education and training for public health and health care professionals to assure awareness of emerging issues and appropriate utilization of new genetic technologies.
2. Work in collaboration with statewide organizations, professional groups and schools of public health and medicine to promote integration of genetics into professional practice.
3. Promote established public health genetics competencies for health care and public health professionals.

Public Education

1. Foster the public's understanding of scientific developments in human genetics and associated ethical, legal and social issues.
2. Initiate and support collaborative public education and training programs that bridge knowledge gaps.

Access to Services

1. Promote access to family-centered, culturally and linguistically appropriate genetics information and counseling, clinical and support services.
2. Reduce financial, geographic, cultural and linguistic barriers to access to genetic services and family support through coordination of services.
3. Promote delivery of community-based genetic services and improve access to quality, cost-effective care.

Information and Referral

1. Develop and maintain a statewide genetics resource database for public education, program planning, policy development and quality assurance related to genetic services.
2. Increase public health capacity for information, referral and technical assistance.

Data Systems

1. Develop integrated data systems to improve data coordination for public health service planning.
2. Strengthen data collection efforts in collaboration with laboratories and comprehensive genetic centers) to develop systems that monitor rates of genetic-related conditions and utilization of genetic technologies.

Public Policy

1. Facilitate development and implementation of public policies pertaining to clinical, ethical, legal and social aspects of genetics services.
2. Maintain communication with professional organizations and foster compliance with clinical and laboratory standards related to genetic medicine, counseling and education.

**Connecticut** Dept. of Health has published the “Genomics Action Plan” in July 2005. The key findings include the following:

1. There is a need for CT DPH to continue to broaden its activities to consider the impact of genomic advances on chronic conditions, infectious diseases, environmental health, and epidemiology.
2. Trends, such as aging population, growing ethnic diversity, childbearing at older ages, and SES disparities, need to be considered when determining future genomic services needs.
3. There is a need to develop a child health information system to assure access to quality health care.
4. There is a need to assure a competent genetics workforce is available to meet the growing demands of genetic testing.
5. b
6. Barriers to access need to be addressed, including availability of trained providers and insurance or reimbursement for services.
7. There is a need to enhance public understanding of genetics and impact of genetics on health.
8. There is a need to coordinate state and federal policy issues and to address community concerns about informed consent, genetic privacy and discrimination.

### **IIIb. Regional Genetics Needs**

In addition to the state needs assessments and planning activities, other groups have engaged in planning activities. One of the activities conducted by the New England Consortium of Metabolic Programs (NECMP) was a needs assessment called *Benchmarks for Fairness* which focused on follow-up of expanded newborn screening. The Benchmarks of Fairness incorporate concepts of equity in risk factors, access and financing, efficiency in the delivery of clinical care and administrative policies, and accountability in terms of how decisions are made, who has access to information, and what methods are used to evaluate the program (Daniels, 2000; Daniels, 2006). A set of nine original benchmarks were adapted for assessing reforms in newborn screening for metabolic disorders.

#### **Benchmarks of Fairness for Health Care Reform (Daniels, 2000)**

Benchmark 1: Intersectoral public health

Benchmark 2: Financial barriers to equitable access

Benchmark 3: Nonfinancial barriers to access

Benchmark 4: Comprehensiveness of benefits and tiering

Benchmark 5: Equitable financing

Benchmark 6: Efficacy, efficiency and quality of care

Benchmark 7: Administrative efficiency

Benchmark 8: Democratic accountability and empowerment

Benchmark 9: Patient and provider autonomy

A manuscript describing a subset of results related to Benchmarks #3 (Non-financial barriers to access) and # 9 (Patient and Provider Autonomy) focused on communication practices in newborn screening and presents some recommendations (Hewlett & Waisbren, 2006). From

this assessment and from additional information obtained from a long-term follow-up study of expanded newborn screening in New England (Waisbren, 2003), 104 infants from minority or non-English speaking families who received a positive newborn screening result since 1999 were identified. The experiences of this group of families have not been previously described and this information will inform our responses regionally. A second result from the Benchmarks assessment suggested that the parent/consumer group for parents of children with PKU and Allied Disorders had little communication with existing professional groups and was not represented on the Boards of other consumer or advocacy groups. A third finding indicated that many young adults with metabolic disorders had been lost to follow-up and others continued to be seen at pediatric centers. Reliable data on the number of individuals followed or lost to follow-up were not available.

**Newborn Screening.** The number of children identified by NBS in New England is increasing rapidly due to expanded NBS programs (Comeau et al, 2004), although each of the 6 states in New England offers a different menu of disorders and services. Laboratory screening for particular disorders yields information on gaps in distribution of available diagnostic and treatment centers for screened positive newborns. Geographic mapping of tertiary care centers that provide treatment for infants affected with the disorders indicates some distribution issues, but does not address level of care, extent of expertise, time availability, whether or not there are genetic counselors trained in handling NBS results, and whether or not there are opportunities for communications between adult and pediatric results.

A need for greater awareness on the part of community physicians has also been identified. A recent survey of 290 pediatricians in Massachusetts (Gennaccaro, Waisbren, & Marsden 2004) indicated that 14% of responders were unaware of expanded NBS and 42% were not comfortable discussing test results with families. The vast majority indicated a need for more information, with 73% recommending postal mailings as the most appropriate format.

**Families and Consumers.** Several evaluations that aim to address identified systems needs as they relate to families have been identified. One is a collaborative study being conducted through the New England Consortium of Metabolic Programs (NECMP) on the Effect of Expanded Newborn Screening for Biochemical Genetic Disorders on Child Outcomes and Parental Stress (Waisbren et al 2003). The goal of this study was to compare identification by expanded screening for metabolic disorders with clinical identification and to assess the impact on families of a false-positive screening result compared with a normal result. Among the findings, it reported increased utilization of ER visits by those with false positive screens compared to the control group. Using a validated instrument (Tluczek et al, 1992; Ciske et al, 2001) for evaluating parent anxiety, comprehension and retention of information showed some acute anxiety relative to the screening result but parents reported they would screen again (Estrella et al, 2000). The New England Newborn Screening Program (NENSP) recent analysis suggests that parents continue to participate in optional NBS but that the model by which genetic counseling is offered has a positive impact on compliance (Comeau et al in press). The New England Newborn Screening Program (NENSP) data show low compliance with genetic counseling offered to families with sickle trait. Further analyses will investigate aspects of the communication processes and health care experiences that may reduce stress in families.

**Education** While recent growth in the field of genetics has produced many changes that could fundamentally alter medical practice, awareness of these changes has not increased with the available information. The recent publication of *Core Competencies in Genetics Essential for All Health-Care Professionals* (NCHPEG, 2003) has not significantly remedied the lack of basic genetics education and knowledge that has been reported among medical practitioners (Wilkinson and Targonski, 2003; Collins, 1997). The general public is less informed about genetic testing than it was in 1990, creating a significant barrier to the effective contribution of genetics to healthcare (Swanbrow, 2003; Rhode Island DOH, 2002; Collins, 1997; Wilkinson and Targonski, 2003). This failure to educate the public occurs against a backdrop of expanded screening options, including expanded NBS (for hearing impairment, cystic fibrosis and additional metabolic disorders) and many other options for genetic screening for adults and entire families (Zytkovicz et al, 2001; Comeau et al, 2004). One recent national study of parent education materials about NBS suggested that materials should be revised to be easier to read and more user-friendly, by lowering the reading difficulty to eighth-grade level and focusing on issues such as layout, illustrations, message, information, and cultural appropriateness. It is important that state NBS programs and organizations work with parents to develop and to evaluate materials to ensure that they are user-friendly (Arnold et al, 2006).

### **Transition to Adult Services.**

Transfer from pediatric to adult health care programs remains a critical issue and will only become more critical as patients who are identified through expanded newborn screening grow older (Schwarz, 2005). Unfortunately, no adult metabolic clinics exist in New England and there is a general deficiency in the number of medical geneticists throughout the region. The target populations for this education initiative are adult primary care providers and teenagers and young adults ages 16-21 years with metabolic disorders identified through newborn screening. Older adults with metabolic disorders who continue to receive their health care at pediatric metabolic centers or who have no medical home may also benefit from this initiative. New England has an adult population (age  $\geq 18$  years) of 13,922,517, of whom 1,353,648 (almost 10%) are uninsured (US Census data, 2000). Based on incidence rates in New England, we estimate that there are approximately 3,393 metabolic patients in this region, of whom at least half (1,700) are over age 18 years. Our own recent reports obtained through the Benchmarks Project from metabolic centers in New England indicate that fewer than 200 patients over age 21 years are regularly followed at metabolic centers. This lack of follow-up may have devastating consequences, particularly for women with PKU who risk bearing children with congenital heart defects, mental retardation and other problems if not treated prior to pregnancy. The problem is two-fold: 1) young adults have difficulty accessing adult metabolic care programs because of a lack of insurance and other psychosocial reasons (Brown, Fernhoff, Waisbren, et al, 2002; Newacheck, Brindis, Cart et al, 1999) and, 2) adult care providers are unfamiliar with metabolic disorders, most of which have traditionally been considered “pediatric” diseases (Lee, 2002). Currently, for adults with metabolic disorders, “crisis management” occurs more frequently than preventive care due to inadequate adherence to medical recommendations and/or intermittent medical follow-up. Patients need a single point of entry into an adult care medical system so that the transition process occurs without care interruption (Brenton, 2000). A patient navigator is needed for those who have linguistic and language barriers (Sarfaty, 2005).

### **II.c. Addressing Planning by State and Region by June 1, 2007.**

The NERC Planning Committee and the new RCC Project Director and Project Coordinator at UNH have reached an agreement with Dr. Tom Brewster, the P.I. of current HRSA cooperative agreement, to request support from carry-forward funds for FY2006 to complete the state and regional needs assessment and plan by June 1, 2007. Specifically, the RCC will facilitate the establishment of planning teams with each state public health department to include public health professionals, consumers/advocates, specialty genetics health care providers, primary care/medical home providers and other key stakeholders. The Project Coordinator will facilitate the team to: a) collect existing needs information, b) identify needs and generate a plan for each state through a one-day facilitated process, and, c) support the writing of a state genetics plan. The Project Director and Project Coordinator will convene a 2-day planning retreat of all state teams, the NERC Advisory Committee, and the NERC Collaborative Council to develop a NE Regional Genetics Plan that will be incorporated into the Goals of the new 5-year cooperative agreement. This is essential to complete the needs assessments and to develop a regional plan. Our aim is to have a shared baseline in all the states and identify specific activities the NERC RCC and Working Groups might undertake to address shared needs. It is recognized that each have differing needs in establishing a shared baseline; and, resources will be necessary to address these needs. We anticipate learning about new or emergent needs by completing the states and regional needs assessments.

### III. METHODOLOGY

Our aim is to improve the health of those with genetic conditions in New England. One model that has been successful in addressing health care systems improvement is that of the National Cystic Fibrosis Foundation (CFF). Recently, the CFF convened Cystic Fibrosis treatment center leaders with national experts on health care quality improvement from Dartmouth Medical School in Hanover, NH and Intermountain Health in Salt Lake City, UT to develop a national quality improvement (QI) effort. The CFF work has been organized by clinical quality improvement and systems improvement principles. They developed a public reporting effort which is part of a comprehensive quality improvement (QI) initiative launched six years ago to accelerate the rate of improvement in quality of care throughout the CF Foundation's care center network. The initiative includes training of care center staff in QI, providing tools to identify and implement best practices, and building stronger partnerships between people with CF, families and their care center clinicians.

The CF quality improvement work is based on the seven “worthy goals” to improve CF care. These goals are:

- To make people with CF and their families full members of the care team;
- To help people with CF achieve normal growth and nutrition status;
- To diagnose respiratory infections early and ensure that the right therapies are received;
- To decrease the spread of germs between people with CF;
- To prevent complications and/or to diagnose and treat them early;
- To provide care regardless of race, age, education or insurance coverage; and
- To support all transplantation and end-of-life care decisions.

We recognize that those with metabolic and other genetic disorders are a more complex group with differing treatment expertise, options, and outcomes. Nevertheless, the complexity of these genetic conditions require a shared conceptual model such as that of the CFF if we are

to succeed in our regional and national collaborative efforts. This health care improvement through learning and collaboration is the “lens” through which we see our success in New England. This “lens” is not explicit, even among our current collaborators, and will require continuous learning by all involved in this important work.

The work of the New England Regional Genetics and Newborn Screening Collaborative (NERC) will be divided among Working Groups, the Collaborative Council, the Regional Collaborative Center at UNH and the Advisory Committee. The work will build on the regional successes of the New England Newborn Screening Program (NENSP). In each state there are key public health partners (e.g., Newborn screening coordinators, MCH, and Title V directors), advocacy groups such as Family Voices, consumers, clinical genetics service providers, federally qualified health centers, and medical home providers. We will establish “state teams” that comprise at least these members based on their particular shared interest in improving programs that support those with genetic and metabolic conditions identified by NBS and later in life. We accept the premise shown in health care improvement research (Batalden and Splaine, 2002 ) that by continuously improving the state programs, health care and support systems, the health of those with genetic conditions will be improved. The NE Region has many national experts in genetics health care who will be key members of NERC. We share the conceptual model that this pooled regional talent will assume the role of leadership, support and facilitation for change and improvement by all state teams. This regional talent pool will populate roles as Working Group leaders and members. Working Groups are organized around themes identified by state needs assessments and to respond to priorities identified in the Program Guidance HRSA-07-016. Each working group leader will be members of the Collaborative Council (CC) whose role is to assure that all state and regional needs are being addressed successfully in the timelines set with the outcomes documented. The Collaborative Council will also serve to address the needs for support to the state teams; they will assure that proper regional “technical assistance” or TA partners are addressing the state team needs. A continuous clinical improvement of health care model (Kasper et al 1992; Ayers et al 2005) will be utilized where appropriate in addressing the needs of each state team. For example, a state may chose to address a problem with lag time between a positive NBS result and the referral to a metabolic center because of geographical distance and lack of transportation. The state team will identify barriers, suggest solutions, test those solutions, measure outcomes and introduce change in systems that will sustain the change. The Regional Collaborative Center (RCC) will serve to assure that the state teams, WG’s, and CC are organized and supported sufficiently with guidance and oversight by the NERC Advisory Committee.

The New England Newborn Screening Program (NENSP) is a model system and sets the stage, in part, for regional systems improvement akin to that of the New England Cystic Fibrosis Treatment Centers (<http://www.dartmouth.edu/~cecs/nnefc/nnefc.html>) . Five of the 6 New England States use the NENSP laboratory located in MA to screen for newborn metabolic, genetic, endocrine and, in some states, infectious diseases. Connecticut has its own NBS program and laboratory. Each New England state health department has a NBS coordinator in place to assure that program performance quality benchmarks are met and comply with state law and regulations. NBS is a private-public partnership among the NBS laboratory, the Department of Public Health and the hospital-medical home-specialist health care providers systems. Each state NBS program has an Advisory Committee (Dr. Moeschler

chairs the NH NBS Advisory Committee) consisting of public health, consumers and family members, primary and specialty health care providers.

To date the NENSP has had many accomplishments that set the stage for continued regional systems improvement and for expanding to genetic conditions identified later in childhood or as adults. Some examples of notable accomplishments include the following:

Health service maps of each state: In cooperation with the departments of health from each of the states of Massachusetts, Maine, Rhode Island, New Hampshire and Vermont, we have established maps of each state onto which NENSP has superimposed client and service data. Some refinement may be in order.

Service database: We have mapped available sub-specialty services. Specialty Centers treating pediatric patients are incorporated into the NENSP database and are mapped within each state's health service areas. NENSP is in the process of mapping adult services and specifying support services available at each center. In addition, NENSP has developed a listing of HRSA funded and HRSA qualified Health centers for all of New England, derived from information available on the HRSA website. Its distinction is that it is functional for mass mailings and communications. A more complete listing of available services is underway.

Case mapping: Using the demographic data available in the NENSP database, NENSP has mapped each confirmed case into the health service areas of the states. NENSP analyzed data for distance between residence and specialty care center.

Case projections: NENSP projected incidences of affected births in those states served by the NENSP for disorders that are not screened in those states. These data were provided to each state's Title V Directors for incorporation into their assessments. Currently NENSP is continuing to map birth cohorts using NENSP generated data. Continuity of mapping high prevalence disorders such as Sickle Cell disease is important to address service needs of more mobile populations such as new immigrant populations. NENSP is adding more disorders to the mapping. Finally, NENSP is working with the CT department of Health in order to map some of their residents' disorders. CT has its own laboratory and NBS delivery system.

NENSP is engaged in *continuous improvement of current practice models* for genetic services relevant to Newborn Screening. Data cleaning for infants characterized with and at risk for galactosemia is complete. Six years of data using the current screening and referral algorithm were characterized. These analyses provide empirical data for providing more accurate risk assessments for each of the kinds of positive screens showing risk for galactosemia.

Improvement of special case issues relative to "border babies" within and beyond Region I as an example of regional collaborative efforts directly impacted by HIPAA. NENSP has hosted three New England regional conference calls in which Privacy Officers, Newborn Screening and Genetics coordinators from each state have participated. These calls provided cross-disciplinary education about the legal and practical issues. It appears from these calls that Massachusetts has the most conservative law and to that end NENSP has held an additional three Massachusetts-specific meetings or conference calls to address current policies and next steps for Massachusetts practices. In addition, with more depth of understanding, the regional group is addressing the priorities of cross border data sharing as well as some of the technical barriers to cross border data sharing.

The Massachusetts Cystic Fibrosis Workgroup and the Regional CF Workgroup developed follow-up guidelines relative to recommendations to be issued this year by the Cystic Fibrosis

Foundation Implementation of Newborn Screening Workshop. Regional workgroups will address the needs of screening in a rural population. NENSP also has established a regional Hemoglobinopathy Workgroup. This workgroup is facilitated by coordinating the meetings through the New England Pediatric Sickle Cell Consortium and is focused on newborn screening issues. The CF Workgroup presented an overview experience of the CF Workgroup as a model for what the Hemoglobinopathy Workgroup might attain.

The NENSP is in the process of optimizing protocols for *long term follow-up* of individuals confirmed to have an inborn error of metabolism, after an abnormal newborn screen or after report of a clinical presentation. This will improve the understanding of the natural spectrum for some of these extremely rare disorders and additionally clarify the benefits of preclinical screening and treatment, if any, in comparison to the natural outcome after a clinical diagnosis. It might also serve as the initial database to aid in setting up a registry for some of these disorders.

Using the NENSP's updated listing of Federally Qualified Health Centers across New England, NENSP developed a survey to assess internet use for genetic queries by staff at these health centers. Questionnaires were faxed to 146 health centers and current response rate is 42%. Responders reported that staff had no capability or time to search for genetic resources.

While we anticipate learning of new needs by completing the states needs assessments and plans in early Spring 2007, we will address the known current themes of needs collectively as well as specific to each state's particular needs: Public Policy and Ethical, Legal and Social Issues; Practice Models and Delivery Systems; Education, Technical Assistance, Dissemination; Genetics Services; and, Public Health Infrastructure.

In addition, we have elected to address specific areas of need: 1) Transitioning from Pediatric to Adult Health Care Services, 2) The Role of the Medical Home in the Support of and Health Care for those with genetic disorders. Laboratory Quality Assurance and Long-term follow-up activities will also be addressed but through the "Additional Priority Activities" and not addressed specifically in this HRSA "Project 2" application.

#### **IV. WORK PLAN**

The NERC Work Plan is organized by an **Activities Approach Logic Model** (see **Attachment 1**) and a detailed Work Plan with specific strategies, activities, timelines, persons responsible, process indicators, expected outcomes and outcomes indicators in contained in the **Work Plan Table** (see **Attachment 1**).

#### **GOAL 1: Establish and maintain a regional Coordinating Center**

The Regional Coordinating Center (RCC) will be established at the University of New Hampshire in Durham at the Institute of Disability in collaboration with the Institute for Health Policy and Practice, both of which are housed in the School of Health and Human Services. Dr. Moeschler will serve as Project Director (PD); Dr. Nisbet, co-PD, and Ms. Philbrick-Schwartz as Project Coordinator. There will be an Advisory Committee consisting of public health, consumers or advocates, genetics specialists and others from each state.

Working Groups will address specific activities. The leaders of each Working Group, together with the PD, will comprise a Collaborative Council. The Collaborative Council will assure all activities are coordinated, collaborative and successful in a timely fashion. The structure resulted from several meetings of the Planning Group in Fall of 2006.

Dr. Moeschler is a Clinical Genetics with 16 years of collaboration experience with the Institute on Disability at UNH. Dr. Nisbet, Director of the IOD/UNH is a national expert on systems and community change activities on behalf of individuals with disabilities. Ms. Schwartz has training in maternal and child public health and is expert in project management, health plan management, negotiation and mediation. Together, this group will lead the RCC and reflect the talents required for collaboration and reflect the interdisciplinary nature of this important work.

**GOAL 2: Collaborate to facilitate access to genetics services, expertise and technology particularly for underserved populations and in rural areas.**

Access to genetics services will be addressed also by the Medical Home and the Transition Working Groups. The Access Working Group, lead by Dr. Moeschler, will collaborate with rural genetics services providers to develop quality improvement projects specific to shared interests and related to issues of access to quality services. Dr. Moeschler has specific expertise in clinical and health care systems improvement as well as clinical genetics. This expertise is unique in our region and will be of use in developing and implementing access improvement activities. The clinical genetics practices in VT, NH and ME serve rural populations with geographic and financial barriers and work with public health to address these barriers. These clinical geneticists have met on two occasions to discuss common interests. The group elected to address the problem of diagnosis and management of patients with intellectual disabilities, a common clinical genetics problem and have implemented a quality improvement project to address this problem. The pilot data have been reviewed and a second iteration has begun. The aim is to improve the diagnostic outcomes of those with global developmental delay or intellectual disabilities for those living in rural areas. The groups status has been presented to the American College of Medical Genetics Special Interest Group on Quality Improvement. The Access Working Group of NERC will continue this activity and extend it to other rural and urban practices addressing those who traditionally experience barriers to services. While the aim is to improve quality of diagnostic services, the secondary benefit will be the increased knowledge and skills of these practices to employ quality improvement principles to their practices. Further, as other practices are added, there may be improvement opportunities regarding other access barriers.

**GOAL 3. Develop effective practice models for adolescents and young adults with genetic conditions who are transitioning from pediatric to adult health care.**

The Transition Work Group will address the needs of state teams and will collaborate closely with the Access WG and Medical Home WG to address barriers to transition to adult health care of those with metabolic and genetic conditions. This WG will serve as the primary resource (Technical Assistance or TA) to develop improvement activities in transition services. Some states (e.g., NH) already have funded Transition projects which may place them ahead in development of others. This WG will work to establish a shared baseline in the region and identify opportunities for improvement in each state.

More specifically, the Transition WG will collaborate with Emerson College faculty and students to develop an “action plan” to continue current efforts to revitalize the Resource Mothers Program (Rohr et al, 2004), a program that in the past received funding to match mothers of children with PKU to women with PKU who are planning a pregnancy or are already pregnant. This program reduced the time women attained metabolic control by four weeks and significantly improved offspring outcome. Approximately 30 women in New England have already been trained to be Resource Mothers, but few are serving to assist women due to lack of funds allocated for this purpose. To create a more sustainable program and expand this program to assist women with other genetic disorders who are struggling with compliance issues, we intend to conduct interviews with a variety of potential stakeholders. The first step will be to meet with leaders of existing prenatal programs and with Dr. Michael Murray, who is developing an adult metabolic program at Brigham and Women’s Hospital in Boston. Once stakeholders are identified, we will convene a series of focus groups that include public health representatives from Maternal and Child Health programs, medical homes, former Resource Mothers, women who have completed a maternal PKU pregnancy and other consumers, representatives from industries that have supported adult treatment initiatives, as well as psychologists, nutritionists, social workers and pre-natal educators. The resulting program or programs will be assessed through measures of adherence to medical recommendations.

The Transition WG proposes to host a NE Conference for Adults with Metabolic and Other Genetic Disorders in 2008. The goal of the conference will be to enhance adherence to medical recommendations in metabolic disorders through: **Social Support, Positive Attitudes toward treatment and Manageability (SAM)**. At the meeting, we will launch the PKU Tool Kit for Transition to Adult Care, which is being jointly developed by the New England Consortium of Metabolic Programs, Emerson College graduate (Aphrodite Veloudaki), and Applied Nutrition, an industry that provides low protein products for PKU and other disorders. This on-line information and educational tool will be adapted for other genetic disorders, since the tool addresses issues of self-advocacy, insurance coverage and attainment of a medical home. The tool kit consists of a compilation primarily of existing materials, many of which were developed through MCH grants.

The National Transition to Adult Care Work Group, now numbering more than 20 people, conducted several conference calls and met at the recent meeting of the Newborn Screening and Genetics Collaboratives. Dr. Waisbren along with Dr. Lou Bartoshesky (NYMAC) co-lead this Work Group. Members of this Work Group represent a much larger group of people involved in regional subcommittees focused on transition to adult care for children with special health care needs, specifically those with metabolic and other genetic disorders. The National Work Group agreed to encourage activities that address the needs and concerns within each region by sharing ideas and resources. The National Work Group also agreed to the following joint endeavors:

- 1) To continue to convene meetings and conference calls for the National Transition to Adult Care Work Group
- 2) To assess preparedness on the part of metabolic clinics and other specialty centers for assisting patients in their transition to adult healthcare. With the help of Patti Hackett, we will adapt and then administer a survey for healthcare providers that was developed by Healthy and Ready to Work (HRTW). Patti Hackett has spearheaded efforts for transition

to adult care for many years and presented an overview of her research and programs during one of the Conference Calls. Completed surveys will be submitted to HRTW for analyses. Regional data will be reviewed. In addition, comparisons will be made with results obtained by HRTW from pediatric practices, family physicians and hospitals. This survey will help identify national and regional needs.

- 3) To identify, adapt and validate a tool (a checklist) that assesses preparedness of patients to manage the transition to adult care. The first grant year, we will work with several organizations, including the Jacksonville Health and Transition Services program (JAXHATS), the Metabolic Program at the University of Washington in Seattle, and HRTW to select the most appropriate currently available instrument. This instrument will then be revised to include questions related to unique issues confronting adolescents and young adults with metabolic and other genetic disorders. The Work Group, in consultation with statisticians and survey specialists, will agree upon methods for validating the instrument and ensuring its reliability. In subsequent years, the survey will be administered to a broad sample of patients. Regional and national data will be analyzed.
- 4) To plan a conference or conferences for adolescents and adults with metabolic and other genetic disorders that will be open to patients from all regions. Before deciding on details, the Work Group will consider issues such as where to hold the conference, which disorders to include, and how to publicize the event. We intend to host the conference or conferences in 2008 so that planning can include all stakeholders and reflect the needs of the collaborating regions.

#### **GOAL 4. Develop effective partnerships to further Medical Home practices in the region.**

“A medical home is a process of care offered by primary care providers in partnership with families of children with special health care needs (CSHCN). Children and families are recognized, welcomed and supported by their community based medical homes.” The American Academy of Pediatrics (2006) describes care from a medical home as: Accessible, Continuous Coordinated, Family-Centered, Comprehensive, Compassionate, Culturally-competent. On the basis of knowledge regarding Medical Home practices for children with special health care needs, it is assumed that the quality of health care and health is improved by successful implementation of such principles in the care of children and youth with genetic conditions. The NERC Medical Home WG will address the systemic change needs in each state and the region. New England is fortunate to have leaders in the development and implementation of Medical Home practices, W. Carl Cooley, MD of NH and Richard C. Antonelli, MD of CT, who will collaborate with Medical Home WG leader, Tom Brewster MD of ME are leaders nationally in this work (letters of support, bio-sketches on file).

The Medical Home WG will work closely with Access WG and Transition WG to improve linkages between clinical genetics and the Medical Home which will be comprised of three distinct yet inter-related areas: 1) Infrastructure, 2) Access, and 3) Healthcare Financing. In partnership with the Primary Care Practice Network developed by Dr. Richard Antonelli and Dr. Carl Cooley in CT, MA and NH as part of their Medical Home Network Initiative, begin to implement projects to: a) improve Medical Home knowledge, and b) implement “transition preparedness” survey in each of 6 pediatric and/ or family medicine practices in the New England region and focusing on the results of state needs assessment, develop and deliver a practice-based training curriculum with associated tools to support transition of youth from pediatric to adult health care in the context of the Medical Home.

**GOAL 5. RCC will serve as the focal point for effective genetics education and dissemination of genetics information.**

The NERC recognizes the importance of creating new knowledge and applying it to the goal of improving the lives of those with genetic conditions. The effective dissemination of information begins with a shared understanding what needs exist and how genetic information is connected to the lives of those living with genetic conditions. NERC has focused its attention on the needs of states in New England and on the needs of key stakeholders. We also strive to have NERC also be a “learning organization”, ie, one in which:

- People feel they’re doing something that matters;
- Everyone is somehow stretching, growing or enhancing her capacity to create;
- People are more intelligent together than they are apart;
- The organization continually becomes more aware of its underlying knowledge base;
- People treat each other as colleagues;
- People feel free to experiment, take risks, and openly assess results.

The Education, Marketing and Dissemination Working Group (EMD WG) led by Dr. Leah Burke of VT will provide stakeholders (consumers, health care providers, policy makers, advocacy groups, educators, and others) with knowledge of genetics and its effects on health in order to contribute to improved health outcomes. The Collaborative has worked closely over the last few years to identify common needs for public health genetics education and strategies for addressing them, and to successfully implement these strategies.

In 2004, the EMD WG performed a needs assessment for the New England states regarding genetics education. Key projects were identified based upon priorities common among most states, and work began on these projects in 2005 via funding from the New England Regional Genetics and Newborn Screening Collaborative HRSA Grant # 1U22MC03959 and in-kind state resources. The Collaborative has monthly conference calls and semi-annual meetings, and has contracted with genetic counselors in the region to provide support and perform project-specific work. Ongoing projects include New England Genetic Resources Directory, Family Health History Outreach, Regional Newborn Screening Brochure in Multiple Languages, Genetics Outreach Project, and development of a New England version of “Understanding Genetics, A Guide for Patients and Professionals” in collaboration with the Genetic Alliance.

The EMD WG will: continue to address the state teams’ educational needs; develop and implement a marketing and dissemination plan; and, evaluate the impact of the EDM WG activities. The EMG WG will not develop new educational products without specific discussions with our HRSA GSG officers. The WG will work closely with the RCC on the development of new website that is organized according to state and regional needs.

**GOAL 6. RCC will demonstrate effective collaborations with other regional and national stakeholders, such as the National Coordinating Center, the National Newborn Screening and Genetics Resource Center, CDC-sponsored Centers of Excellence for Birth Defects Prevention Research, CDC Newborn Screening Branch, NICHD, AAP,**

**University Centers of Excellence in Disability (UCEDD's), MCH Leadership Education in Neurodevelopment Disability Education programs (LEND programs),etc.**

The NERC intends to “promote a new model of relationships” within the region and with national partners, as has been formulated by Peter M. Senge (1994): 1) share all relevant information with all partners in a form that all can understand, 2) share credit, for collaboration and shared authority cannot exist when credit is hoarded, 3) reward and recognize honesty and openness, 4) promote and reward partnering, and 5) tend to persons’ perceptions of relationships. At least one specific collaborative activity for each stakeholder will be identified by the Collaborative Council. Collaboration with key stakeholders is essential for the success of the NERC. We will continuously identify new partners with which to collaborate to the extent that the collaborations enhance our capacity to improve the lives of those with genetic conditions in New England.

**GOAL 7. RC will examine the relevant public policy and ethical, legal and social issues affecting individuals with genetic conditions, their families, and health care providers and educators.**

In a recent commentary, Botkin et al (2006), stated:

*“A central problem in the debate over expanded NBS is the lack of information on the efficacy of current NBS programs and the lack of sufficient research about the treatment of the conditions and the impact of screening to clearly justify policy decisions about which tests should be performed and/or disclosed to parents... In the future, DNA-based technology will permit analysis for an even larger number of conditions and genetic susceptibilities using dried blood spots. Given these capabilities, a fundamental problem is whether to manage tests for different conditions on the multiplex panel as separate tests or as a single test package. If programs choose only to disclose results on conditions that clearly meet established criteria, then results on the other conditions, and the potential benefits flowing from those results, will be withheld from families and care providers. On the other hand, offering results on a large number of conditions for which there is limited or no evidence of benefit to affected children may cause harm to some children and families and is likely to be a poor use of scarce resources. This is a legitimate dilemma for which we can offer no easy solution.”*

The expansion of newborn screening tests in recent years has moved so quickly that the public policy and ELSI issues have not been fully articulated, discussed and collectively resolved. It is essential that a lively discussion of the implications of expanded newborn screening be central to the NERC activities in the region and nationally. We plan to collaborate with the Mountain States (MOSTGENE) and other regions to create an open discussion of the relevant issues. Dr. Jeffrey Botkin, UUTAH, has agreed to collaborate on our efforts.

In addition, state teams and state Newborn Screening Advisory Committees have had to address decisions on expansion with the existing body of knowledge regarding the principles of newborn screening first articulated by the WHO in the 1960s (Alexander & van Dyck, 2006). The PELSI WG will collaborate to assist state teams in addressing their needs.

**GOAL 8. The RC will utilize a small project program to accomplish unanticipated and innovative activities that emerge within the region.**

To provide the flexibility of NERC to respond to emerging needs, innovate ideas and unfinished state and regional genetics plans, we have proposed a innovate grants program for each year of the cooperative agreement. A review process is proposed modeled after that of the Heartland Region and we have requested \$100,000 in year one to be utilized to fund projects up to \$30,000 each. In addition, the Education, Dissemination and Marketing Work Group has requested \$15,000 to support small projects prioritized for consumers and advocacy groups not to exceed \$5,000 which will be administered by NERGG, Inc.

**GOAL 9. The RC will provide opportunities to increase discussions with school systems and state special education leaders to improve access and support to educational services in the least restrictive environment.**

This is a new opportunity for collaboration in our region, particularly given the expertise of the RCC. Children with metabolic or genetic conditions often have special health care needs that impact their education. We will approach the issue through collaboration with other groups that have a responsibility to improve special education services: the North East Regional Resource Center on Special Education located within WestEd; the Parent Training and Information Centers (most have a direct relationship with Family Voices), the University Centers for Excellence on Developmental Disabilities (many have an expanded mission to serve all children and adults with disabilities), the MCHB Leadership Education in Neuro-Developmental Disabilities (LEND) network, and the state Special Education Directors Association. We are fortunate to already have linkages with these groups through current and previous state and regional demonstration, training, technical assistance, research, and evaluation efforts.

One of the first tasks will be to identify and articulate the nature of the educational issues that children with metabolic and other genetic conditions have. Many of the children do not qualify for special education services because they do not have an educational disability. Instead they are served under Section 504 of the Rehabilitation Act of 1973 provisions. Based on our experiences, this often creates a level of uncertainty related to what services and supports and/or accommodations are required. We intend to help clarify these issues, and collaborate with groups who have both expertise and experience with these issues.

Furthermore, there is a need to educate physicians, genetic counselors, and other health care professionals who have frequent contact with children with genetic conditions and their families. They too need information on best practices; as well as tools to support families to advocate for their children to be educated in the least restrictive environment and to have access to the regular education curriculum. We are particularly interested in graduation outcomes and post-secondary education attendance. These outcomes combined with performance on state-wide assessment tests will provide a useful baseline for states to understand academic achievement levels of children with genetic conditions.

**GOAL 10. The RC will complete both quantitative and qualitative evaluations of processes and outcomes of all goals, activities and projects undertaken by the NERC.**

The creation of the annual and final evaluation report will be based on the results of a four-year evaluation that will focus on two key areas. One, what information about the project's

progress will enable project staff to ensure successful completion of project goals. Two, what occurred as a result of the grant funds (i.e., to what extent were all the objectives met within the states and the region?). Additionally, we will track the number of study findings from projects that are published in peer reviewed journals to address the **MCHB Performance Measure 03**.

Evaluation efforts will be fully integrated into all project activities and guided by core project assumptions of collaboration, partnership, state and regional improvement and sustainability. Making the commitment to engage in collaborative processes requires that evaluators and all project participants be inclusive, open to dialogue, partnership oriented and committed to reasoned deliberation. Fetterman and his colleagues (1996) suggest collaboration is a key strategy of an “empowerment evaluation” approach. Collaboration serves to foster empowerment goals such as quality assurance, system improvement and consumer involvement and satisfaction. Many of these ideas are drawn from the growing literature describing collaborative and participatory evaluation (Sohng 1995; Stringer 1999). Making the commitment to engage in collaborative processes requires that evaluators and all project participants be inclusive, open to dialogue, partnership oriented, and committed to reasoned deliberation. The ultimate goal is that the evaluation efforts will support the development, implementation, and sustainability of collaborative efforts that are directed at overall improvement in the NE states’ and the region’s ability to provide quality genetic, newborn screening, medical home, follow-up, laboratory, educational, and transition services and supports to children with genetic conditions and their families.

When considering varying evaluation approaches for their fit with project assumptions and principles, the strengths and weaknesses of both traditional and non-traditional models were considered. Traditional models have alienated many populations, including elders, individuals with disabilities, their parents and family members, because efforts are typically conducted in an isolated manner and have little or no impact on current project efforts. Important concerns in conducting significant sustainable contemporary evaluation are: 1) consumer satisfaction, 2) greater meaning and relevancy, 3) higher quality data, and 4) more effective utilization of findings (Hagner 2002). Another major factor considered when selecting the project’s evaluation model was that the goal was not to just assess and understand the effectiveness of the *NERC* model, but to formulate, implement, and track solutions that will support sustainable and transferable systemic changes to the states and region.

A variety of measures will be reviewed by the Advisory Committee to assess the project, including process (e.g., have specific tasks been accomplished in accordance with expectations?), and outcome (e.g., children transition to quality adult care) measures. Data will be collected through currently existing structures within participating organizations as well as data collection forms created by the project’s evaluator, Dr. Antal (with input from the Advisory Council, the NCC, and Collaborative Council). Frequency of data collection activities will be based on availability of data sets and the requirement for annual performance reports. Qualitative data from Advisory Council, Collaborative Council, Working Group, and state team meetings, service logs, and checklists will be analyzed using content and pattern analysis techniques to evaluate the process of implementing the *NERC* goals and objectives. State and regional level data that meets confidentiality and privacy standards related to each of priorities will be collected and descriptive analyses and correlational/multiple regression techniques will be used to interpret these data. A combination of software packages, including

Filemaker Pro (database design and management), NVIVO (qualitative analysis), Excel (graphical representation), and SPSS (statistical analysis) will be used to conduct analysis and update staff throughout the project.

**Evaluation Focus #1: Provision of an Information Feedback Loop to Advisory Committee, RCC Staff, Collaborative Council, and Working Groups**

In accordance with the first area, evaluation work will seek to assist NERC’s staff and collaborators in maintaining a clear and consistent understanding of the project’s status concerning the completion of primary goals, levels of participation in the project by stakeholders, as well as recurring concerns and important new developments that may influence the direction of the project. Evaluation strategies for internal management focus on two approaches. First, *formative evaluation* techniques will be used to evaluate whether the project activities are being conducted as proposed and planned (if not, why not and what changes can be made to revise the plan without jeopardizing the integrity of the project). This approach will include a comprehensive examination of project records and files, determining if deadlines are being met in addressing each research activity. It is in this phase of the evaluation that the *process* of the project is examined to understand many of the environmental factors that impact on success of the project, and improve the project continuously. Each month, the Management Team will complete the *Progress Evaluation Chart* together in a staff meeting. Activities associated with each goal and objective will be reviewed as assigned code of 1-7, depending on their status:

<b>1. Completed as planned</b>	<b>4. In progress - unsatisfactory</b>
<b>2. In progress - satisfactory</b>	<b>5. Initiation of activity deferred</b>
<b>3. Completed - deviated substantially from plans</b>	<b>6. Activity abandoned</b>
	<b>7. Not scheduled to initiate this month</b>

Any activity receiving a code of #3, #4, #5, or #6 will be discussed with the RCC Management Team in order to remediate ineffective implementation, suggest new activities and timelines or justify elimination/delay of the activity entirely. These monthly assessments will be used to improve the performance of the project throughout its duration.

During the course of NERC’s implementation, the Project Evaluator with support by the Project Directors and Project Coordinator will be responsible for writing quarterly reports which will provide updates to project staff and collaborators on the following areas: a) project status relative to overall project timeline and goals; b) participation among project staff and collaborators; c) barriers/facilitators to achieving goals and objectives; and d) recommended changes to project implementation.

As a part of this process, Project Evaluator, Dr. Antal and Project Coordinator, Amy Schwartz will maintain records of minutes from all project meetings as well as issues identified during the course of the project’s implementation. Furthermore, given the complex nature of the NERC, it is vitally important to ensure that a wide variety of voices and perspectives are reflected in both the creation of research tools as well as in the interpretation and dissemination of results. To this end, meeting minutes will track major issues identified as well as levels of participation by key stakeholder groups (HRSA Project Officers, Advisory

Committee, Collaborative Council, and Working Groups). In addition, project participants will be asked to complete a brief anonymous web-based annual survey in order to address the following areas: a) satisfaction with their level of involvement in the project; b) clarity of their role in the project; c) extent to which key issues and concerns of the groups they represent are included in project implementation; and d) suggestions for project improvement.

### **Evaluation Focus #2: Achievement of Goals and Objectives, Summative Evaluation**

In assessing whether key goals of the grant and project were met, we will be using a range of performance measures drawn from best practice literature, including measures cited within the originating RFP and recommended measures from the National Coordinating Center. During the first year of the NERC, the Working Groups will review and identify specific outcome measures related to their content area. These outcome measures will be reviewed by the Advisory Committee and approved. Additionally, during the last six months of the project, Advisory Committee members will participate in a focus group to discuss the extent to which the vision and plan for the NERC was achieved. Some of the focus group questions will be tied to each project goal.

In order to determine the extent to which project services met the needs of the individual states as well as the region as a whole and contributed to the achievement of goals, all participating stakeholders will be asked to complete a brief web-based survey at the end of the project concerning the use and applicability of each of the major components of the project. Key areas will address: a) satisfaction with the efforts to pursue state and regional goals, b) ratings on the utility of each component area in helping the states to achieve their individual goals, c) additional supports needed to achieve state and regional goals, and d) ff additional technical support was sought during the project, and if so, were they satisfied with the technical support received.

The final evaluation report will also include a summative report which addresses the areas cited above, discusses the barriers/facilitators which had an influence on NERC project activities, identifies evidence of change, and provides recommendations for a next cycle of state and regional improvement.

## **V. RESOLUTION OF CHALLENGES**

Coordination has been defined as the managing of dependencies between activities. Kraut and Streeter (1995) combined the various definitions from other coordination researchers and formally defined coordination as "the direction of individuals' efforts toward achieving common and explicitly recognized goals and the integration or linking together of different parts of an organization to accomplish a collective set of tasks." Because the RCC has multiple goals, objectives, and organizations involved in meeting shared outcomes, attention must be paid to project management, communication, and evaluation.

**Challenges.** We have experienced and identified a number of challenges to coordination and collaboration with multi-site teams as well a solutions. We identified these challenges by interviewing key individuals involved with the RCC, reviewed historical documents

including extensive communication with the HRSA project officers, and reviewed the literature on collaboration and effective multi-site teams.

CHALLENGES	SOLUTIONS
<b>Multi-site and individual diversity and experience</b>	
Lack of contact among team members	We plan to have an annual regional planning meeting; an annual meeting, monthly Collaborative Council and Work Group meetings, and bi-yearly Advisory Committee meetings. The Management Team will meet monthly. In addition, we will make available web-based meeting rooms through the web-site and sponsor monthly “webinars” given by RCC members.
Physical separation	Unfortunately, we are unable to change the geography of the region but the location of the NERC at UNH in Durham provides an ideal location for the multiple organizations and individuals involved. The New England Center, a Kellogg funded conference center, was established at UNH to serve the purpose of promoting regional collaboration. Key stakeholders will also be encouraged to meet during other national meetings, e.g., ACMG and NSCG.
Lack of trust	There are several new players in the RCC; in particular the IOD and the IHPP at UNH. We intend to address issues of collaboration and trust during the first annual Regional Planning Meeting through a facilitated discussion.
Personal work style differences	Every collaborative project includes individuals with different work styles. We intend to address work style differences and how to support one another in our work during the first regional planning meeting.
Different backgrounds of team members/ New team formation	There are a variety of backgrounds among members. Different professional and personal backgrounds can enrich a project if used wisely. UNH is fortunate to have one of the foremost experiential education facilities in the country which we will utilize during the planning retreat to help to create trust and understanding. Additionally, there are new teams that will be formed both at the project and state level. Each team will need to be supported by the project to assure that systems change (state level teams) and project activities are accomplished.
History	The NERC has had its own unique history which should be acknowledged. During discussions with key stakeholders, it is clear that individuals are prepared to move forward and create a new foundation for genetics and newborn screening in NE.
Lack of meaningful engagement of consumers of services.	The NERC requires that 25% of the Advisory Committee include consumers of services. In addition, we are asking that each workgroup include at least one consumer. Family Voices as well as several other “disability specific” groups will be invited to participate in the regional planning effort as well as the annual meeting. These groups in each of the states will be queried as to their participation.
Lack of attention to	We will be reaching out to the Community Health Care Centers, e.g., Bi-state Primary Care or the Rural Health Collaborative; as well as

the underserved.	organizations that represent traditionally under-presented groups. They will be invited to be part of the Advisory Council as well as participants in annual meeting. Each state team will include a rural health partner.
<b>Communication across organizations, management structures, and individuals</b>	
Loss of ad hoc communication to identify and resolve issues	Informal contacts over time build trust and assist in identifying “issues” in real time rather than as part of a formal communication structure. We intend to develop a specific web-based space for individuals to identify issues (and potential solutions) in real time.
Communication differences or preferences.	There are several individuals involved in the Project who are not frequent e-mail users or do not have appropriate technology, including universal design. Amy Schwartz, Project Coordinator will query all members of the Advisory Committee and Council about their preferred communication methods and modes. We will attempt to accommodate these differences as well as assure the website and communication use principles of universal design. Amy Schwartz MPH has specific training in negotiation from the Kennedy School at Harvard. She will use these skills in her role as Project Coordinator
Not realizing there is a need to communicate.	We intend to establish predictable communication and reporting requirements. Each subcontractor and/or service provider will be required to prepare brief quarterly updates on objectives and activities. Additionally, at the monthly Council meetings, we will have a specific agenda item that address this issue.
Lack of time to initiate contact or communication.	The NERC recognizes that time is an issue for everyone, but believes that providing clear guidelines and timelines, and efficient rather than cumbersome communication strategies that we can improve communication and work towards strategies that are mutually beneficial.
Lack of knowledge and use of appropriate decision-making strategies.	Decision-making is not problem solving. We intend to use a decision-making matrix that clearly distinguishes between authoritarian, sub-group, majority, and consensus strategies and how and when they are used. These strategies will be particularly important with the Collaborative Council and the Working Groups.
Lack of systematic use of web-based and/or distance technologies for communication and learning.	The NERC will be investing in the development of a new website that will serve multiple needs including dissemination of knowledge, communication, identification of resources, and project management. We will have the resources of UNH to assist in utilizing up-to-date distance communication technologies.
<b>Project Management</b>	
Involvement of multiple stakeholders with different incentives and agendas; lack of clarity about roles	Any project that involves the distribution and management of resources, financial and human, requires clear expectations. The NERC has developed a Memorandum of Understanding for the contractors and service providers ((Attachment 4). We will develop, collaboratively, with the Advisory Committee, Collaborative Council, RCC Management Team, and Working Groups similar documents

and responsibilities	that outline expectations and responsibilities during the first quarter, year 1.
Duplication of processes	The challenge for the NERC is to establish shared process but do so in the way that does not centralize all functions and bureaucratize the project. Quarterly reports will be submitted on common forms. A centralized web-site will use the best of individual efforts. Data collection processes will be streamlined and, when appropriate shared across states and the region. Conference planning will be conducted by the NERC but the Workgroups will be managed by the WG Leaders as will be the state teams.
Lack of appropriate advisory and management structures	The NERC has established multiple management structures including a Management Team responsible for assuring that project goals, objectives, and outcomes are reached; and financial processes are consistent with federal requirements. The RRC has not had this structure in the past. To assure participation of states, we will support an Advisory Committee designed to provide input into the overall project, guide the development of specific activities, and participate in the evaluation process to assure we are meeting our responsibilities. The Collaborative Council assures that the Chairs of the Workgroups have regular communication and input into the development of new goals and activities.
Inattention to issues of sustainability	Sustainability will be addressed as a regular agenda item within each of the Management Structures. As part of the evaluation process, the issue of sustainability of activities will be addressed. If work is being conducted that can not be sustained, a problem solving process will be used to decide how to get additional resources to make it sustainable.
Lack of shared outcomes and indicators	The NERC will develop a set of shared baselines for each of the states and a set of agreed upon outcome activities for each of the major project area, e.g., improving special education. Because some of the states do not have comprehensive state teams, we will work to support teams that can help to identify data sources and shared baselines.
Lack of use of shared evaluation processes and indicators.	The NERC will use an empowerment evaluation (Fetterman, 2000) strategy to engage and improve project performance. This strategy requires ongoing assessment of what is working and what is not and allows teams and participants collectively to identify new solutions and processes. Dr. Antal is skilled in this area of evaluation and will support the project to use this approach.

**VI. EVALUATION AND TECHNICAL ASSISTANCE CAPACITY**

The New England Regional Genetics and Newborn Screening Collaborative (NERC) intends to establish the Regional Collaborative Center (RCC) at the University of New Hampshire. The RCC consists of individuals with extensive experience in project management, evaluation and technical assistance. Each individual and their organizational capacity are described below:

**Regional Collaborative Center Staff and Management Team**

John B. Moeschler MD, MS Project Director, and Chair of the Policy, Ethical, Legal and Social Issues Work Group, and member of the Access WG is the Director of Clinical Genetics and Professor of Pediatrics at Dartmouth Medical School. He received his MD from the University of Nebraska and completed his Fellowship in Developmental Pediatrics at the University of Washington. In 1984, he was Board Certified in Medical Genetics. Dr. Moeschler recently (2005) received his M.S. in Health Care Improvement from the Center for Evaluative Clinical Studies at Dartmouth. He chairs the Advisory Committee on Newborn Screening for the State of New Hampshire; and is PI on a CDC funded Birth Defects Registry. He is also the PI on the MCH funded NH Leadership Education in Neuro-developmental Disabilities (LEND) program which is a collaborative project between Dartmouth Medical School, Department of Pediatrics; and the UNH School of Health and Human Services including the IOD. He has worked extensively on the island of Guam and in Micronesia providing technical assistance to MCH and Special Education to develop clinical and follow-up systems for children with disabilities. Dr. Moeschler participated in the Dartmouth Moral Philosophy project funded by ELSI (NHGRI) that resulted in the publication of the book “Morality and the New Genetics”. He has published extensively in the field of clinical genetic and developmental disabilities; and recently published a paper in Pediatrics entitled: *The Diagnostic Evaluation of the Child with Mental Retardation or Global Developmental Delays* in his role as a member of the AAP Committee on Genetics.

Jan Nisbet Ph.D., Project Co- Director and Chair, Working Group on Improving Special Education, is the founding Director of the Institute on Disability, A University Center for Excellence on Disability at the University of New Hampshire. She received her Ph.D. from the University of Wisconsin-Madison. She was worked professionally as a pediatric physical therapist, special educators, augmentative communication consultant, and faculty member. Over the past 20 years, the Institute on Disability has been the recipient of approximately 90 million dollars in federal and state grants directed at improving the social and economic lives of children and adults with disabilities. The IOD served as a National Program Office for the Robert Wood Johnson Foundation and provided technical assistance to over 20 states. Additionally, she served as PI on a national housing initiative for people with disabilities that involved 30 states. In NH, she and IOD staff work closely with state and community agencies to support children and adults with disabilities in inclusive environments. She has served as PI on numerous grants and contracts focused on improving educational, employment, family support and community systems. She is the author of numerous publications and chapters, and editor of several books focused on community services and supports. The IOD has a staff of 40 individuals, 10 of which have disabilities or are family members of individuals with disabilities. Dr. Nisbet presents and provides technical assistance on developing community based services and supports nationally and internationally. Her most recent article published in the Journal of Community Development is entitled “*Community-Centered Strategies for Individuals with Disabilities*”. She also co-authored a book entitled “*The Inclusion Facilitators Guide*” published in 2006. Dr. Nisbet is a tenured associate professor in the Department of Education at the University of New Hampshire

Amy Philbrick Schwartz MPH, Project Coordinator, is currently the Associate Director of the Institute on Health Policy and Practice at UNH. She received her MPH from the University of Hawaii-Honolulu with a specialization in Maternal and Child Health and a minor in disabilities studies. In her role, she manages core staff of professional analysts, consultants and project associates and oversees daily operations including budget and interdisciplinary project administration. In addition she negotiates grants and contracts with State government and federal agencies. She also serves as the Director of the Health

Information Security and Privacy Collaboration, is staff to Governor's Task Force on Healthy New Hampshire, Health Care Purchasing subcommittee. She has served as the lead for Medicaid cost containment initiatives and program evaluation projects including provider reimbursement, long term care planning, and case management for patients with chronic illness and program operations. As a policy analyst for IHPP, Ms. Schwartz was the primary liaison to the State of New Hampshire Medicaid Division and Healthy Kids Corporation. Prior to working at UNH, she was Vice President of Provider Network Management for the Neighborhood Health Plan in Boston/Harvard Pilgrim where she lead, managed and directed the activities of the Provider Network Management division including staff management, contracting, financial analysis, network development, provider relations, provider communication, provider database management, and credentialing staff for a 110,000 member Medicaid HMO. Additionally, she analyzed provider contractual arrangements and prioritized re-contracting efforts to ensure fiscal solvency and administrative ease; and was responsible for negotiating financial agreements and maintaining relationships with key constituents in provider community. She is a highly respected manager and has extensive experience providing technical assistance to state agencies, community health care centers, and health care organizations.

Peter Antal, Ph.D., Project Evaluator, is currently the Research Director at the IOD at UNH and a clinical faculty member in the Department of Health Policy and Management. He received his Ph.D. in Urban Affairs and Public Policy from the University of Delaware. He recently completed a comprehensive evaluation using empowerment evaluation methods in the assessment of the Littleton Model Community project where his key responsibilities included quantitative analysis of surveys, qualitative analysis of stakeholder interviews and focus groups, reporting on and dissemination of project "lessons learned." For the past two years he has been collecting data for New Hampshire's first state of the state report on individuals with disabilities. Additionally, he initiated New Hampshire's first state-wide study on the transportation characteristics and needs of individuals with disabilities and the elderly as well as the broader public. Prior to working at UNH he was the Research Director at the Children's Alliance of New Hampshire where he collaborated with staff to produce and disseminate special reports on child well being in New Hampshire, including the *Kids Count New Hampshire 2003 Data Book*. Peter has also initiated and led the development of the New Hampshire Census Research Group, a unique coalition of 50 researchers (representing fields of housing, economic security, health, social services, planning, and education) working together to enhance the quality and effectiveness of New Hampshire's research and evaluation community. He also led the development of a multi-state research project assessing family economic success in rural areas of northern New England. He was also the evaluation coordinator for the Wilmington, Delaware Health Start Empowerment Evaluation Center at the University of Delaware as well as several other evaluation projects involving home visiting and parents as teachers. Peter recently published as part of the State of the State Series, the first report on special education services in NH.

### **NERC Working Group Chairs and Members**

Susan Waisbren, Ph.D., Chair of the NERC Transition to Adulthood Working Group, is an Associate Professor of Psychology in the Department of Psychiatry at Children's Hospital in Boston. She received her Ph.D. from the University of California, Berkley in Clinical Psychology. Since 1978 she has served as a psychologist in the Clinic for Inborn Errors of Metabolism and Phenylketonuria at the Developmental Evaluation Clinic and the Division of Genetics, Children's Hospital, Boston. Susan has been the recipient of numerous national research awards and since 1984 has served as the Co-Director, and since 1991, Coordinator of Psychology Follow-up for the NIH/NICHD funded Eastern

Contributing Center, Maternal PKU Collaborative Study. Since 1999, she has been PI for the New England Metabolic Resource Center, the Maternal PKU Resource Mothers Program, Expanded Newborn Screening for Metabolic Disorders Study, Director of the New England Consortium of Metabolic Programs, and the PI on the current NERC subcontract on metabolic disorders. She is the author of numerous publications on psychosocial issues facing families and children who have metabolic disorders. She was the lead author on a recently published article entitled: “*Effect of expanded newborn screening for biochemical genetic disorders on child outcomes and parental stress*”, which appeared in JAMA.

Anne Comeau, Ph.D., Chair of the NERC Follow-Up Working Group, is Deputy Director of the New England Newborn Screening Program and Associate Professor at University of Massachusetts Medical School. She received her Ph.D. in Biology from Brandeis University and conducted post-doctoral studies at the Harvard School of Public Health. Since 1991 she has directed the Molecular Development and Diagnostics Laboratory which is part of New England Regional Newborn Screening Program. Since 2004, she has served as a co-investigator and project leaders of the NERRC. She has been an invited expert and speaker to CDC, NIH, the Hastings Center, and HRSA on topics related to screening and genetic conditions. She is the author of numerous publications on newborn screening, surveillance, public health laboratory protocols, and quality assurance. One of her most recent publications that will appear in In Press (a publication from the Hastings Center) is entitled: “*Population-based research within a public health service: Two models for common rule compliance in the Massachusetts newborn screening program*”.

Roger Eaton, Ph.D., Chair of the NERC Laboratory Quality Working Group, is the Director of the New England Newborn Screening Program (NENSP) at the University of Massachusetts Medical School and serves as Chief of Serology of the NENSP. Roger received his Ph.D. from SUNY Buffalo in Microbiology and completed post-doctoral studies at Harvard Medical School in Immunology. He has provided leadership to HRSA in areas such as the challenges associated with newborn screening related to state policies and procedures”. He is a member of the HRSA sponsored National Laboratory Workgroup for Newborn Screening Program Evaluation and Assessment Scheme (PEAS) Development; and was an invited expert to a workgroup co-sponsored by APHL, CDC, HRSA, NNSRG to develop strategy to definitively determine the need for routine second specimens for effective newborn screening in the US. He is internationally renowned and recently delivered the plenary address on Developments in Newborn Screening in Mexico. Dr. Eaton is the author of over 20 publications. One of his most recent publications entitled: “*Newborn Screening: New Developments in a Proven Field*” was published in Clinical Laboratory Science.

Joanna Fanos, Ph.D., Member of the NERC Follow-up Working Group with Anne Comeau, Ph.D. and Susan Waisbren, Ph.D. is a Research Assistant Professor in the Department of Pediatrics at Dartmouth Medical School. She received her Ph.D. in Human Development and Aging at the University of California, San Francisco. She is the founder and current Director of the Sibling Center at the California Pacific Medical Center. She has held several positions as lecturer in the area of adolescent development at Berkeley and the University of California. Joanna was also a visiting research in the Medical Genetics Branch of the National Human Genome Research Institute. She has consulted and provided technical assistance in the area of sibling loss and genetics across the country and with other regional coordinating centers. She currently serves on the Newborn Screening Advisory Committee at the University of Utah; and serves on the Cystic Fibrosis Studies Consortium, Ethical, Legal, and Social Implications Branch of the Human Genome Program. She has published extensively in the field in the areas of genetic testing, cystic fibrosis, sibling loss and grieving. One of

her most recent publications entitled “*Attitudes toward Prenatal Testing and Screening for Fragile X*” was published in Genetics in Medicine.

Leah Weyerts Burke, M.D., Chair of the NERC Education, Dissemination and Marketing Work Group, is a Professor of Pediatrics and Internal Medicine at the University of Vermont College of Medicine. Dr. Burke received her M.D. at the University of North Carolina School of Medicine degree in 1987. She completed her pediatric residency at the University of California at San Diego in 1990, followed by a fellowship in Dysmorphology and Clinical Genetics also at the University of California at San Diego, completing her training in 1992. She joined the faculty at the University of Vermont College of Medicine as an Associate Professor in 2000, and was promoted to Professor of Pediatrics and Internal Medicine in 2006. Dr. Burke is board certified in Pediatrics and Clinical Genetics. Since coming to UVM, she has been active in the designing and implementation of the new medical school curriculum known as the Vermont Integrated Curriculum (VIC). Dr. Burke serves as the director of the Vermont Regional Genetics Center and is the clinical advisor for the Vermont Department of Health Metabolic Clinic, the geneticist for the Vermont Craniofacial Team, and the medical consultant for the Vermont Newborn Screening Program. She also serves as the genetics advisor for a CDC funded project to develop a birth information surveillance system for Vermont. Dr. Burke serves on the Executive Council of the Section on Genetics and Birth Defects of the American Academy of Pediatrics and is the current senior Co-Director of the New England Regional Genetics Group. She is the author of numerous chapters and articles related to genetics, malformations, and screening. Her most recent publication in the Journal of Inheritable Medical Disorders is entitled “*New England Consortium: A model for medical evaluation of expanded newborn screening with tandem mass spectrometry*”.

Tom Brewster, M.D., Chair of the Medical Home Working Group, has served as the Project Director of the NERC since 2004 and is the Director of the Metabolism Program at the Barbara Bush Children’s Hospital at Maine Medical Center in Portland. Tom received his M.D. from the University of Nebraska and completed fellowships in developmental pediatrics, and metabolism and genetics at Children’s Hospital, Boston. From 1978-1997, he was the Director of Medical Genetics at the Foundation for Blood Research in Scarborough, Maine. He served as the Co-Director of the New England Genetics Group (NERG) from 1992-1994. He is a member of the American College of Medical Genetics Special Interest Group on Public Health, and is a consultant for the Maine Newborn Screening Program. He has numerous publications; and co-author of several articles with Dr. Susan Waisbren on biochemical genetic disorders and screening. He recently was a co-author on an article entitled “*Newborn screening compared to clinical identification of biochemical genetic disorders*” which was published in the Journal of Inheritable Metabolic Disorders.

## **VII. ORGANIZATIONAL INFORMATION**

The Institute on Disability and the New Hampshire Institute for Health Policy and Practice at the University of New Hampshire (UNH) are the applicant organizations, in collaboration with the Dartmouth-Hitchcock Medical Center Department of Pediatrics, Division of Medical Genetics. Each of these New Hampshire-based organizations, experienced in grant management and project administration, has on-site staff and administrative structures which facilitate effective collaboration across all six New England states. Centrally located and the intersection of three major interstate highway systems, New Hampshire has a regional airport in Manchester, 35 minutes from the University of New Hampshire and one hour from Dartmouth Hitchcock Medical Center, making

regional travel convenient and comparably inexpensive. UNH is also the site of an Amtrak station between Boston and Portland, Maine.

**The University of New Hampshire** at Durham (UNH) is the flagship research university within the University System of New Hampshire. The schools and colleges blend general education with state-of-the-art programs in 100 academic disciplines with more than 13,500 students enrolled in degree programs. The 610 graduate faculty members and 2,500 graduate students at UNH work together to develop new theoretical and empirical knowledge, design innovative methods and technologies to discover and disseminate that knowledge, and engage in undergraduate and graduate state-of-the-art teaching. As a land, sea, and space-grant research university, UNH maintains a strong concern for the social applications of the work, seeking to apply new understandings and new discoveries to the challenges that face our state, region, nation, and world. UNH is recognized internationally for its research in Family Violence, Global Climate Change, Marine Sciences, Disabilities, and Outdoor Education. In the fiscal year 2006, UNH received over \$128 million in awards from competitive proposals, an increase of 18.4% over the previous year. Grants are administered through the UNH Office of Sponsored Research, which has staff experienced in state, federal and private foundation award management.

This project will be housed within the **School of Health and Human Services (SHHS)**. The SHHS houses the allied health training graduate education programs in occupational therapy, nursing, speech language pathology, social work, family studies, recreation management, and health management and policy. SHHS has 379 graduate students, more than 56 faculty members, and is the fastest-growing school at UNH.

**The Institute on Disability/University Center for Excellence on Disability at UNH**, a partner in this project, is a nationally recognized leader promoting full community participation for people with disabilities and their families through professional development, research, model demonstration, technical assistance, leadership development, systems changes, and policy analysis. As part of a 55-program UCEDD (University Centers for Excellence on Developmental Disabilities) network, the IOD works to enhance knowledge, utilization, and dissemination to improve the lives of people with developmental disabilities. The IOD has a faculty and staff of 55, and is the only UCEDD in the country that has a formal partnership among a state university, a medical school (Dartmouth Medical School) and a law school (Franklin Pierce Center for Health, Law and Ethics). The IOD was founded in 1987 in a partnership with the NH Department of Education and the NH Department of Health and Human Services to bridge the work of the state and the university. In 2003 the IOD was chosen to receive the President's Award from the National Down Syndrome Congress "for its years of sustained, cutting edge leadership in the disability field and particularly for its commitment to including parents and self-advocates as equal partners. Currently the IOD administers nearly \$5 million in state, private and federal funds for projects benefiting infants, toddlers, children, and adults with developmental and other disabilities and their families in the areas of: early care and education, early identification, family and consumer leadership training, interdisciplinary professional leadership education, access to the general curriculum for students with significant disabilities, inclusive education, positive behavioral approaches, assistive technology, personalized budgets for consumers, systems change, co-worker support, self-determination, managed care, autism, and accessible and affordable transportation and housing for people with disabilities. The IOD has offices in Durham, Concord and Manchester, NH; and print material, videos, and research papers for national distribution. The IOD is known for its ability to bring NH based research and demonstration projects

to national scale. The Director of the IOD, Jan Nisbet, PhD, will provide general project over site and direction as well as leadership in the area of special education to the NERC.

**The New Hampshire Institute for Health Policy and Practice (NHIHPP)** at UNH was established as a formal alliance between the State of New Hampshire Department of Health and Human Services Division of Medicaid Business and Policy, the University of New Hampshire and Dartmouth Medical School. The mission of the Institute is to create a more cost effective and responsive community health system, and focuses on a population-based approach to health improvement.

The founding Director of the NHIHPP, Ned Helms, a former Commissioner of Health and Human Services in New Hampshire, was hired in February of 2001. Since that time, the Institute has received State, Federal and Foundation grants to undertake projects across a range of Health Policy areas. The Institute is actively engaged in applied, community-based research projects to include:

1. Applied Research and management to create an Inventory of Public Health Information and best practices resources for the State.
2. The creation of curricula and educational programs for the Public Health workforce within the New Hampshire at the State and local level.
3. The creation of a first in the nation consumer-focused web site, in conjunction with the NH Insurance Department on cost and quality information relating to frequently performed in and out-patient procedures.
4. Qualitative and Quantitative research on the extent of informal care giving for the frail elderly in the State, including the development of a family caregivers guide distributed throughout the state.
5. Policy development and evaluation for the creation of a state wide network for provision of information and support for families and individuals through Aging and Disability Resource Centers.
6. Support for a comprehensive Health Initiative led by the Governor of the State to address the Health and Health Care System through the New Hampshire Health Initiative, a decade long effort to address Quality, Health Promotion, and Information and Cost Analysis.
7. Leadership of a state wide effort to examine the evolution and promulgation of Health Information Technology and Exchange within the State.
8. Undertaking in cooperation with Columbia University a multi-year research project on the effects of arsenic in the drinking water of school districts within New Hampshire.
9. Ongoing assistance to the State Medicaid program in the area of contracting, information analysis and management, and program innovation and development.

The Institute also serves as a convener of health researchers in Northern New England to focus on areas of common concern relating to the analysis of Discharge and Claims information, the development of Health Information Technology and Exchange, and the engagement of the broader population in the shaping and evolution of the Health and Health Care Systems of Northern New England.

The NHIHPP has staff experienced and trained in the fields of public health, maternal and child health, epidemiology, survey design and evaluation. The Institute is independently funded through grants and contracts and is housed on the main campus of UNH, Durham. Amy Philbrick Schwartz,

MPH and Associate Director of the NHIHPP will serve as project manager for the Regional I Genetics and Newborn Screening Collaborative. Both the NHIHPP and the IOD report directly and are accountable to the Dean, School of Health and Human Services (**Attachment 4**, Letter of Support). The Directors of both Institutes serve on the Executive Council of the School.

**The Dartmouth-Hitchcock Medical Center, Department of Pediatrics, Clinical Genetics Program** provides diagnosis, treatment, genetic testing and genetic counseling for individuals known or suspected of having inherited medical problems. The Department of Pediatrics at Dartmouth Medical School is the academic and clinical department for all faculty providing health care services for children at the Children's Hospital at Dartmouth and throughout the Hitchcock Clinic system in NH and Vermont. The nation's fourth oldest medical school, it melds the resources of a world-class medical center with the breadth of an Ivy League institution and builds on more than two centuries of scholarship to shape exceptional leaders with the skills and confidence to excel. Dartmouth Medical School (DMS) promotes discovery and dissemination of knowledge to harness advances in biomedical research and health care assessment to benefit society and cure disease. Investigators collaborate across the departments of Dartmouth Medical School and Dartmouth College to understand basic mechanisms and translate their insights into new treatments and technologies. An array of multidisciplinary programs and centers at the interface of science and medicine builds bridges for cutting edge research that leads to novel therapies and their scientific, outcomes-based evaluation. DMS is on the front lines with notable contributions in such areas as cancer, cell and molecular biology, genetics, infectious conditions, immunology, bioethics, neurosciences, public health and medical outcomes. The Director of the Clinical Genetics Program, John B. Moeschler, M.D., is a Professor in the Department of Pediatrics in the Section of Genetics and Child Development and is a developmental pediatrician and medical geneticist. Dr. Moeschler will serve as the Project Director for the Regional I Genetics and Newborn Screening Collaborative.

### **Administrative Structure**

The New England Regional Genetics and Newborn Screening Collaborative (NERC) will establish a Regional Collaborative Center (RCC) at UNH which will provide leadership, facilitate coordination, manage all aspects of the NERC, and provide fiscal administration. Each of these three organizations: the IOD, NHIPP, and the DHMC Department of Pediatrics, Clinical Genetics Program, will contribute to the management of the NERC. NERC is organized in two ways: 1) each state has designated membership on the Advisory Committee from public health, genetic services as well as a consumer representative; and 2) NERC will establish eight Working Groups to include ethical, legal and social issues; improving special education, medical home, education, dissemination and marketing; transition to adulthood, laboratory quality and follow-up. The Working Groups will be chaired by content area experts and each will work with all of the New England States (see **Attachment 5** Project Organizational Chart). The NERC has worked to assure that there is leadership from each of the states in the region to assure that state level needs are being addressed as well as Regional. NERC Advisory Committee: The 18 member Advisory Committee (3 members from each state; one member will be a consumer of services) will develop a mission statement and formalize the expectations of the content-area working groups. The Advisory Committee will assure that there are members from traditionally underrepresented groups as well as individuals from rural areas. Additional members may be added to address specific initiatives such as improving access to high

quality special education services. The Committee will review the work of the Collaborative Council and Working Groups and determine, with the management team, effective dissemination of materials and work products to maximize benefit to providers and consumers. Additionally they will participate in the annual and final evaluation surveys to determine project effectiveness (See **Attachment 5**).

The Regional Collaborating Center (RCC) Management Team will: 1) convene and staff the Advisory Committee, Collaborative Council and provide support to the Working Groups; 2) facilitate meetings, define roles of group members and maximize participatory engagement, 3) encourage cross-state input and support, 4) augment participation of working and advisory groups as appropriate, 5) conduct formative and summative evaluation as well as to promote empowerment evaluation strategies, 6) revise activities, promote project innovation in cooperation with the HRSA project officer as necessary in order to maximize project success and achieve project goals, and 7) prepare reports and publish results. They will meet weekly to monitor progress and assure that goals and objectives are being completed in a timely and high quality fashion. The management team will consist of John Moeschler, Jan Nisbet and Amy Philbrick-Schwartz.

The Collaborative Council is comprised of the chairs of each of the Working Groups. It will meet monthly and be chaired by Dr. Moeschler. The purpose of the Council is to provide ongoing input into the NERC and to develop collaborative and coordinated activities across the multiple content areas.

Working Groups: The eight Working Groups will serve as a focal point for identified and specific areas and each will be chaired by a respected New England expert. The work of the groups will address the specific goals and objectives of the NERC and improve access and capacity in clinical and public health systems for families and consumers across the region, and to advance best practices in the region.

State Teams: Each state will convene teams to identify state needs and to collaborate to identify regional resources to assist in addressing needs, including participating in state and regional planning. The State Teams will include, at minimum, the State NBS coordinator, consumer, advocacy group (e.g., Family Voices), specialty genetics provider, Medical Home provider, and Federally qualified health center representative.

(N.B. All citations on file; available on request)