

TENNESSEE GENETICS PLAN

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PURPOSE OF THE PLAN:

The purpose of the statewide genetics plan is to integrate the Newborn Genetic Screening (NBS) Program and other genetic services into the Information Technology (IT) Plan to assure an adequate public health infrastructure and to coordinate genetic services with all other programs for children with special health care needs (CSHCN).

This plan proposes to 1) enhance, expand and improve the State Newborn Screening (NBS) program by integrating NBS and Newborn Hearing Screening (NHS) with all aspects of Maternal and Child Health Services (MCH) for children with special health care needs (CSHCN) and with early intervention services and 2) enhance, expand and integrate the Tennessee Department of Health (TDH), Information Technology (IT) program called Children's Information Tennessee (CIT) to monitor access to care and track the health status of children identified by NBS and NHS programs and by other programs for children with genetic conditions.

The activities proposed in this plan are based on the recommendations of the **Newborn Screening Task Force** (Pediatrics 106:25, pp 383-427, 2000) and of the Agenda for Healthy People 2010. Tennessee has a **Newborn Screening (NBS)** system with adequate public health infrastructure and a highly coordinated team of public health staff, professionals at the genetic and sickle cell centers, and pediatric endocrinologists who interact with the hospital staff that collects the samples and with the primary care providers (PCPs) (medical home) and specialists who provide medical care to assure early diagnosis and treatment of all infants with presumptive positive results. The NBS program is also coordinated with the NHS program and with the CIT program.

The NBS program was expanded and strengthened by increasing the number of tests performed on all newborns and by integrating the NBS and NHS services with follow-up and early intervention services in order to create a continuum of care from diagnosis and treatment to coordination with other MCH Title V community based system of services for CSHCN and with PCPs (medical home) and specialists, family support, and early intervention services.

The primary outcomes of the Agenda for **Healthy People 2010 are addressed**. The mission statement of the Consumer Involvement Subcommittee organized as part of our Genetics Planning Grant is to ensure that families are educated, empowered, and engaged as satisfied decision-making partners in advocating for and receiving health and related services for a child who has or is at risk for chronic physical, developmental, behavioral, or emotional conditions (CSHCN).

GENETIC SERVICES IN TENNESSEE:

INTRODUCTION:

Tennessee was one of the first ten states that received Federal MCH funding from the National Genetics Disease Act (Title XI of the Public Health Service Act) in 1978. In 1985 the state passed genetic legislation funding the continuation and expansion of the genetic services developed with federal funds. This legislation also created a **Genetic Advisory Committee (GAC)** to advise the Commissioner of the Department of Health in the development, expansion, and maintenance of Regional Genetics and Sickle Cell Centers and to develop standards for statewide genetic services. The Commissioner of Health appoints the members of this Committee. The committee is comprised of one representative from each

Regional Genetics and Sickle Cell Center and at least two members-at-large. The Chief Medical Officer of the state or an appropriate designee chairs the committee. The GAC, must by law, meet on site at least once per year and has been meeting twice a year since its inception in 1985.

TENNESSEE DEPARTMENT OF HEALTH (TDH):

The **TDH** is a branch of state government with a Commissioner appointed by the Governor. TDH has divided the state into 13 health regions that administer services in the 95 county health departments. Seven of the regions are comprised of rural counties, and six are metropolitan counties under the jurisdiction of metropolitan governments. The counties in the rural regions are part of the state's administrative system, whereas the six metropolitan counties are part of county administrations. The local health departments participate in the follow-up of the Newborn Genetic Screening Program by performing the re-screening, and help in locating infants who need confirmatory evaluation and/or follow-up plus providing the prescribed special formulas through WIC. The Section of MCH includes the **Newborn Genetic Screening and Genetic Services Programs** and the **CSS program**. The CSS Program has served as the medical home for children with special health care needs and is now more involved in care coordination if the child has a primary care physician assigned by the TennCare MCO or supported by insurance. This program is also involved in NHS follow-up. TDH is organized into five bureaus and CIT reports directly to the Assistant Commissioner for BHI who reports directly to the Commissioner of Health. This provides CIT with optimal access to both programmatic and information technology resources. BHI has two divisions: Policy Planning and Assessment (PPA) and IT. Vital Records, Health Statistics, Health Research fall under the division of PPA. One of the key functions of BHI is to provide timely, appropriate, secure access to all health data sets. BHI maintains the central data repositories and works closely with program areas on research, evaluation and policy development.

NEWBORN SCREENING (NBS) PROGRAM:

This program is mandated by law and is responsible for the laboratory services performing the newborn screening tests. The program is under the administration of the Section of Maternal and Child Health of the Department of Health. The screening for **PKU** started in 1968, **hypothyroidism** was added in 1980, newborn screening for **sickle cell anemia and other hemoglobinopathies** began in 1988, **galactosemia** was added in 1992, and **congenital adrenal hyperplasia** started in the year 2000.

The testing methodology and protocols are clearly outlined in the Newborn Screening Procedure Manual and the laboratory follows quality control measures, and rules and regulations governing the program. This laboratory services all children born in the State of Tennessee (over 79,400 resident births and over 82,000 total births each year). Follow-up of positive or abnormal screening results is performed according to protocols approved for each disorder.

The staff of the State program is responsible for providing the results of the screening test to the PCPs (medical home) and reports all presumptive positives for PKU and galactosemia to three Genetic/Metabolic Centers located in Memphis, Nashville, and Knoxville. Presumptive positives for hemoglobinopathies are reported to the Sickle Cell Centers, and those for hypothyroidism and congenital adrenal hyperplasia are reported to regional pediatric endocrinologists. **Confirmatory diagnostic testing** is conducted as follows: **PKU** – at one of three Genetic/Metabolic Centers;

Galactosemia – at a reference laboratory recommended by the Metabolic Center and approved by TDH; **Congenital Hypothyroidism and Congenital Adrenal Hyperplasia** in a laboratory licensed by the state; **Hemoglobinopathy** – at the laboratory of Meharry Comprehensive Sickle Cell Center.

In November of 2002, the GAC recommended to the Commissioner of the TDH that the NBS program be expanded to include four additional tests. The Commissioner submitted a formal request to the Governor to expand the NBS program and the Tennessee Legislature approved the request. Screening for **Biotinidase** will start by July of 2003 and testing for **Medium-chain Acyl CoA dehydrogenase Deficiency (MCAD)**, **Homocystinuria**, and **Maple Syrup Urine Disease (MSUD)** will begin as soon as the Tandem Mass Spectrometry (TMS) equipment is installed and training and quality assurance are completed. In May of 2003, the GAC recommended additional screenings for other genetic disorders detected by TMS. The experience of other states such as California and Massachusetts will be reviewed and national recommendations from the Expert Group of the HRSA funded contract to the American College of Medical Genetics titled “Standardization of Outcomes and Guidelines for NBS Progress” will be followed.

NBS TREATMENT AND FOLLOW-UP SERVICES:

Treatment and follow-up services for PKU and galactosemia are performed by three Genetic Centers / Metabolic Centers located in Memphis, Nashville, and Knoxville. Treatment and follow-up for sickle cell disease is provided at the Sickle Cell Centers: Meharry Comprehensive Sickle Cell Center in Nashville, at the St. Jude Comprehensive Sickle Center in Memphis and at two Satellite Sickle Cell Centers in East Tennessee located in Chattanooga and Knoxville. The follow-up of screening for congenital hypothyroidism and congenital adrenal hyperplasia is coordinated by a network of pediatric endocrinologists who act as consultants to the PCPs (medical home) treating these infants.

The Genetic/Metabolic and Sickle Cell Centers confirm the diagnosis and treat the infants screened by the NBS program and provide information to the parents on the specific disorder and on support groups. They also coordinate services with the medical homes and with case managers from TDH Children Special Services (CSS) and Tennessee Early Intervention Services (TEIS). Recommendations are also made for other early intervention and rehabilitation services (PT, OT, speech therapy, etc.).

NEWBORN HEARING SCREENING (NHS) PROGRAM:

This program is not mandated by law but in 2003 about 98% of the TN birth population has access to voluntary hearing screening.

The **NHS** Program started in 1997 when the state began collecting information from birthing hospitals and other professionals. A task force was formed to address screening methods, education, legislation, assessment and early intervention. By March 1999, twenty-eight hospitals with 54% of the birth population had universal NHS equipment available and eleven others were screening high-risk infants and those infants in Newborn Intensive Care Units (NICU). Beginning in September 2000, hearing-screening results were voluntarily reported on the Newborn Screening lab slip creating an initial surveillance system for Universal NHS. The NHS was linked to the NBS in 2001 and results of both NHS and NBS are entered into the State Information Technology (IT) system called

Children's Information Tennessee (CIT). In April 2003, 82 of the 89 birthing facilities were conducting universal newborn hearing screenings. This indicated that 98% of the TN birth population had access to hearing screening. By July 2003, all but two small birthing hospitals will provide hearing screening.

OTHER GENETIC SERVICES:

Consultations for other genetic disorders are provided by three comprehensive genetic centers located in Memphis, Nashville and Knoxville. These Centers offer clinical genetic consultations, specialized laboratory services (clinical cytogenetics, biochemical and molecular genetics), out reach clinics and professional and community education. Two satellite centers located in Chattanooga and Johnson City offer clinical and cytogenetic services. The state also has a network of Sickle Cell Centers.

These centers receive funding from the TDH and participate in the GAC. Close collaboration between the centers and the TDH has developed a network of genetic services that offer the latest technology in the field to rural and urban populations of the state. The last decade has seen many advances in genetics, most notably the sequencing of the human genome by the "The Human Genome Project". In the next few years medical genetics will become a very important aspect of all fields in Medicine and Public Health. These centers will play a very important role in the development of new advances in both fields of medicine.

GENETIC CENTERS

The Department of Medical Genetics (Developmental and Genetic Center) **University of Tennessee Medical Center, Knoxville**

Program Director: Carmen B. Lozzio, MD, FACMG

Clinical Genetic Services: The patients and families are scheduled at the Center and at Outreach Genetic Clinics located at public health departments and local pediatrician's offices in ten counties of East Tennessee. In-patient consultations are available as needed (24hrs/7days/all year) at UT Medical Center and area hospitals.

These services include general genetics, prenatal counseling, risk counseling for breast cancer and other hereditary cancers, and counseling and diagnostic services for neurologic disorders (Huntington disease and hereditary ataxias). Diagnosis, treatment and follow-up services are provided to infants with metabolic disorders such as PKU and Galactosemia, identified through the State Newborn Screening program.

Outreach Clinics: [Huntsville](#); [Kingston](#); [LaFollette](#); [Loudon](#); [Madisonville](#); [Morristown](#); [New Tazewell](#); [Newport](#); [Oak Ridge](#); [Sevierville](#)

Genetic Testing: The laboratory services offered by this Center are:

Clinical Cytogenetic and Molecular Cytogenetic Laboratory:

Carmen B. Lozzio, MD, FACMG, Director. This laboratory has one MD clinical cytogeneticist and seven cytogenetic technologists. A total of 2,000 chromosome analyses are done per year including samples of amniotic cells, CVS, blood, bone marrow, products of conception, tissues and solid tumors. Molecular cytogenetic studies (FISH) are performed in prenatal samples, blood, bone marrows, and solid tumors. This laboratory also offers new techniques such as M-FISH and multi-telomere studies.

Biochemical Genetics Laboratory: Karla J. Matteson, PhD, FACMG, and Director. This laboratory performs all the phenylalanine and tyrosine analyses for diagnosis and monitoring of the treatment of infants, children and adults with PKU identified by newborn genetic screening program. In addition the laboratory provides amino acid, organic acid and mucopolysaccharide analyses for diagnostic evaluation of infants and children from surrounding hospitals and at the Center. The laboratory analyzes approximately 2000 samples yearly. The laboratory also performs over 4000 yearly Maternal Serum Profile testing for pregnant women.

Molecular Genetics Testing Laboratory: Karla J. Matteson, PhD, FACMG, Director. This laboratory provides diagnostic and predisposition testing for a variety of Mendelian Disorders. Among the specific tests performed are Factor V Leiden, prothrombin, MTHFR, PAI, Prader Willi/Angelman, Fragile X, cystic fibrosis, Friedreich Ataxia, Huntington Disorder, multiple SCA types and other neurological disorders. The laboratory analyzes over 2000 samples annually.

Collaborative projects: Drs. Lozzio and Matteson participate in clinical and research activities with groups within the University of Tennessee Medical Center and with other academic research groups and private industry. These efforts involve projects to identify genetic influences on the development of cancer, genetic polymorphism predisposing to cardiovascular disease and the development of new mutation detection instrumentation.

Future direction for expansion includes:

Common complex disorders: counseling and testing for cardiovascular disorders and thrombosis, hypertension and diabetes. Clinical applications of research discoveries.

Autism, ADHD, and other behavioral disorders: Research counseling and development of laboratory tests, as more information becomes available on the genetics of these disorders and on the role of neurotransmitters.

Molecular "CHIP" DNA microarrays: Participation in research to develop new techniques and apply them to the diagnosis of genetic disorders.

For more information, check the website:

www.utmedicalcenter.org/genetics

The Division of Medical Genetics (Department of Pediatrics) Vanderbilt University School of Medicine.

Program Director: John A. Phillips, III, MD, FACMG

Clinical Genetic Services: General genetic clinics are available at Vanderbilt University Medical Center (VUMC) and in outreach clinics at public health departments in Middle Tennessee.

In-patient consultations are available 24 hours a day, 7 days a week at Vanderbilt Hospital and at area hospitals upon request.

Specialty genetic services at VUMC include metabolic, prenatal, hemostasis-thrombosis, cystic fibrosis, Down syndrome, familial cancer and predictive testing [for neurological conditions including Huntington disease and hereditary ataxias and familial primary pulmonary hypertension (FPPH)] clinics. Enzyme replacement therapy is provided for a variety of metabolic conditions including Gaucher, Fabry, MPS 1 and 1-S through the Lysosomal Storage Disease Center. Prenatal and specialty counseling is also provided two days per week at Baptist Hospital and at outreach clinics.

Outreach Clinics: [Columbia](#); [Cookeville](#), Clarksville (Fall 2003)

Genetic Testing: This center has the following genetic laboratory services.

Biochemical testing: The Clinical Biochemistry Laboratory, based in the Pathology department, provides plasma and urine amino acid studies which are interpreted by Medical Genetics faculty. Other testing includes maternal serum screening (first and second trimester). Medical Genetics faculty will also assist in interpretation of tandem mass spectrometry results at the Tennessee State New Born Screening Laboratory (fall 2003).

Molecular Diagnostics: The Molecular Diagnostics Laboratory, directed by Dr. Cindy Vnencak-Jones, offers a variety of tests, including DNA analysis for carbamyl phosphate synthetase 1(CPS1)and medium chain acyl CoA dehydrogenase deficiencies, cystic fibrosis, fragile X syndrome, Huntington disease, factor V Leiden, prothrombin, MTHFR and BMP2 (risk factor for FPPH) gene mutations, T and B cell and BCR rearrangements that are associated with lymphomas and chronic granulocytic leukemia and detection of engraftment after bone marrow transplantations for malignancies.

Collaborative Research: Research currently being conducted in the Division of Medical Genetics (Department of Pediatrics) involves collaborators within the Vanderbilt community, the United States (academic and private industry) and the international community. Areas of focus include causes and treatment of collagen disorders such as spondyloepiphyseal dysplasia tarda, and urea cycle disorders, the molecular genetics of nitric oxide metabolism, glutathione and growth hormone deficiency, hemochromatosis, angioedema, autism, mitochondrial disease, multiple lipoma, carotid artery tumor and multiple keloid syndromes, dyslexia, familial obesity, persistent pulmonary hypertension of the newborn and following cardiac surgery, PPH and idiopathic pulmonary fibrosis and genetic counseling issues in predictive testing for complex diseases. In addition, the Medical Genetics Division has taken a leadership role in the Vanderbilt Clinical Research Center Genetic Initiative

Future directions:

1. Expansion of existing services. The Division of Medical Genetics (Department of Pediatrics) has had a 60% increase in patients seen over the last four years (15% growth rate per year). Increased demand for genetic services is also anticipated due to the addition of 20 tests to the Tennessee Newborn Screening Panel and increased awareness of genetics and advances in genetic research and technology, especially related to cancer and complex diseases.

2. Research in developmental biology: As the genome project matures, current estimates suggest that 30,000 to 40,000 human genes will be identified. Studies of the contributions of each of these genes to growth, development and disease will constitute a major focus of research into the next millennium.

For more information, check the center's web site:
www.mc.vanderbilt.edu/vch/interior.php?mid=178

University of Tennessee, Memphis - Genetic Center, at the Department of Pediatrics, Division of Medical Genetics, and Department of Obstetrics-Gynecology, Division of Reproductive Genetics.

Program Director: Jewell C. Ward, MD, PhD, FACMG

Clinical Genetic Services: The division has three full-time ABMG Certified Clinical Geneticists (Dr. R. Wilroy and Dr. E. Pivnick, in addition to Dr. Ward); they hold 8 General Genetics Clinics per month. Specialty Genetic Clinics include Inborn Errors of Metabolism Clinics (2/month) and a Genetic Neurofibromatosis Clinics (2/mon). Outreach General Clinics (2 /month). Clinical Geneticists provide immediate genetic evaluations to 7 birthing hospitals in Shelby, and are on the consulting staff of an additional 2 hospitals. Services include diagnostic genetic evaluations of individuals with dysmorphology, genetic counseling for at risk family situations (multiple fetal wastage, family history of genetic disorders, reproductive risks due to exposures) and presymptomatic genetic evaluations. The division has affiliation with the Department of OB/GYN, ABMG Certified Reproductive Clinical Geneticist, who holds weekly prenatal diagnostic services, including advanced ultrasonography, amniocentesis and CVS procedures. The center provides services to citizens in over 25 counties in W. Tennessee, as well as citizens in E. Arkansas, N. Mississippi, N. Alabama, and S. Missouri. This Center, in conjunction with the U.T. Boling Center for Developmental Disabilities, has provided follow-up diagnostic and nutritional treatment services for the designated W. Tennessee catchment area for presumed positives of hyperphenylalaninemia and galactosemia. Similar services are provided to E. Arkansas and N. U.T. Boling Center Metabolic Nutritionists provide specialized services to dietary-treated patients attending the IEM Clinic, and Dr. Ward monitors the PKU patients and their families at the U.T. Boling Center weekly. Mississippi. Genetic associates include an ABMG Certified Genetic Counselor, and two Genetic Nurses, providing additional family services to patients.

Outreach Clinics: Jackson and Arlington Developmental Center, Arlington, TN

Genetic Testing: This Center has the following genetic laboratories:

Clinical Cytogenetic and Molecular Cytogenetic Laboratory:

Aviradan Tharapel, PhD, FACMG Director. The laboratory performs chromosome analyses in samples of amniotic fluid, CVS, and cultured tissues, from LeBonheur Children's Hospital, birthing hospitals from the Shelby County area, private physicians and as referral from regional laboratories. Molecular cytogenetic studies are performed to detect microdeletion syndromes, translocations, and markers. Research is performed on new molecular cytogenetic techniques

Inborn Errors of Metabolism Laboratory:

Jewell C. Ward, MD, PhD, FACMG, Director. The laboratory provides a battery of screening tests for inborn errors of metabolism (amino acids, carbohydrate, storage and organic acid disorders) and maintains affiliations with other academic IEM specialty laboratories for the definitive evaluation, diagnosis and confirmation of IEM. The lab participates in the on-site monitoring program in the U.T. Boling Center for Developmental Disabilities for PKU patients in W. Tennessee, N. Mississippi and E. Arkansas. **Research interests**, Dr. Ward is primarily involved in clinical research on metabolic patients as pertains to collaborative protocols in inborn errors of metabolism, such as treatment of storage disorders.

Molecular Genetics Laboratory:

Vickie Park, PhD, FACMG, Director. **Research Interests:** NF1, and genetic predisposition in minority populations to hepatitis.

Education:

All faculty and genetic associates participate in didactic and clinical training to medical students, pediatric, medicine-pediatric and family practice residents, fellows, faculty, primary care providers, public health professionals, and the public.

Future expansion of Genetic Services includes:

Enhancement of existing presymptomatic genetic counseling services, participate in initiatives for treatment of storage disorders (Gaucher disease ongoing for 10 years, addition of Fabry disease, MPS-I, and GSD-II collaboration). Continue to forge linkages with non-pediatric departments on campus (Medicine, Family Practice), the basic science molecular resource centers, and to expand Genetic Service Training of primary care providers. Increasingly develop strategies for the transfer of information to primary care providers.

Genetic Center at the University of Tennessee College of Medicine at Chattanooga and T. C. Thompson Children's Hospital

Program Director: Cathy A. Stevens, MD

Clinical Services: The Genetics Program at the Chattanooga Unit of the University of Tennessee College of Medicine and T.C. Thompson Children's Hospital provides comprehensive genetic services to Chattanooga and the surrounding counties. Genetic services are provided by outpatient genetic clinics as well as inpatient consultative services for patients with a variety of birth defects, metabolic disorders, developmental impairment and prenatal counseling. Outreach clinics are held at the Whitfield County Health Department in Dalton, Georgia and at the Orange Grove Center which provide outpatient programs for developmentally handicapped children and adults. Genetic counseling is also provided through multidisciplinary clinics including the Craniofacial Clinic and the Muscular Dystrophy Association Clinic. We have recently established a Genetics Oncology Clinic and incorporated it into the comprehensive oncology services available through Erlanger Medical Center. Inpatient consultations are performed frequently in the Newborn Intensive Care Unit, pediatric inpatient units, labor & delivery unit and the adult inpatient services. Examinations of fetuses are conducted for genetic evaluation and counseling regarding fetal loss.

Clinical Cytogenetic Laboratory

Program Director: Mary C. Phelan, PhD,

This laboratory has one PhD clinical cytogeneticist and four cytogenetic technologists. The laboratory performs chromosome analyses in samples of amniotic cells, CVS, blood and bone marrow. Molecular cytogenetic studies are also performed

Genetic Center at East Tennessee State University ,Johnson City

Program Director: Jack Rary, PhD,

This center provides clinical genetics and cytogenetic services for patients from the Upper East Tennessee area

For more information, contact Dr. Jack Rary at (423) 439-8541, rary@etsu.edu

LOCATION OF GENETIC CENTERS AND OUTREACH CLINICS

The following map from Gene Clinics show the distribution of clinical genetic services in the state:

TENNESSEE



SICKLE CELL CENTERS

Meharry Comprehensive Sickle Cell Center

Director: Steven M. Wolff, MD

This Center was developed in 1972 and was initially funded by the state of Tennessee. This program received federal funding for 10 years (until 1998) and presently receives support from state funds only. This Center is a multi-disciplinary effort, encompassing many departments of Meharry Medical College, and its main teaching hospital the Nashville General Hospital at Meharry. It includes both Clinical and Basic Science Departments to implement and provide the best possible care for individuals with sickle cell disease. The Center also cooperates with extra mural facilities, such as: Vanderbilt University Medical Center and, the Lentz Health Center of the Metro-Davidson County Health Department.

Goals and Objectives: The goals of the Meharry Medical College Comprehensive Sickle Cell Center are: 1) to provide access to comprehensive and quality services for sickle cell patients and their families. 2) To provide a vehicle for clinical and basic research with a primary focus on sickle cell disease and other hemoglobinopathies. 3) To focus resources, facilities, and manpower in order to maximize the Center's efforts and 4) To provide adequate experiences for the teaching of health and allied health professionals and scientists, 5) Provide confirmatory testing for hemoglobinopathies for the Statewide Newborn Nursery Program. 6) Provide genetic counseling and education.

Service Population: During the period from 1972 through 1998, data has been collected on more than 500,000 individuals who were screened by the Meharry Laboratory. The majority (91%) of these individuals proved to have normal hemoglobin either of the adult or fetal type. More than 80% of the remainder have sickle cell trait. Those with sickle cell disease formed 2% of the screened population. Individuals with rare types of hemoglobin such as β -thalassemia, α -thalassemia trait, SC Harlem, and HPHF ranged from 0.006% to 0.3% of the population. The Meharry Comprehensive Sickle Cell Center's Laboratory continues to provide the state with the confirmation laboratory support for its Newborn Screening Program. The annual reports on the number of samples processed by the laboratory unit of the Comprehensive Sickle Cell Center documents that on average more than 6,000 infant samples are processed annually. It is estimated that one of every ten non-white infants born will have sickle cell trait (Hb AS). Based on previous annual data of 16,805 non-white births, they estimate that between 1,600 and 1,700 infants will be born with the Hb S trait each year in Tennessee. This means that their parents and usually their siblings will need evaluations, followed by counseling services. Meharry's Comprehensive Sickle Cell Center provides the necessary genetic counseling services, as well as, the training of public health nurses in Middle and East Tennessee.

Hemoglobinopathy Detection and Reference Laboratory: Meharry's Comprehensive Sickle Cell Center maintains the Hemoglobinopathy Laboratory for the State of Tennessee. State-of-the-art laboratory equipment is utilized presently for the statewide sickle cell program. The laboratory also provides confirmation testing for the state of Tennessee Newborn Screening Program. The Meharry Hemoglobinopathy Detection Laboratory currently performs about 13,000 screening tests for hemoglobinopathies annually. Over the past two decades, over 500,000 screening tests have been performed on the at-risk populations of our state; including those on infants, children and adults. As the Hemoglobinopathy Reference Laboratory for the State of Tennessee, it currently provides confirmatory diagnoses for newborn screening specimens. The Meharry Laboratory is licensed by the State of Tennessee, and for the purposes of quality control, participates in the proficiency testing

programs of the College of American Pathologists. The laboratory has had a greater than 90% proficiency rating for the past 10 years.

The Sickle Cell Center and Meharry Medical College: The Sickle Cell Center is a health care delivery unit of the School of Medicine of Meharry Medical College. Its research investigators have had appointments in the departments of Biochemistry, Pathology, Pediatrics, Medicine, the Graduate School, the School of Allied Health Professions, and the Center for Nutrition and the Clinical Research Center. Meharry provides the Sickle Cell Center with all of the support that is essential to make the center a viable health care entity for sickle cell patients. The Office of Research Support, the Computer Center and the Office of Grants and Contracts all play a major role in assisting the center in implementing its stated objectives.

Community Relationships: The Center has had ongoing relationships with other agencies in its catchment area over the years. Such agencies are: The United Ways of Clarksville and Middle Tennessee, The Counsel of Community Services, Tennessee Department of Health, Metro-Nashville Health Department, Planned Parenthood of Nashville and the Tennessee State Legislature.

St. Jude Comprehensive Sickle Cell Center

Director: Winfred Wang, MD

The Pediatric Hematology Center of Memphis and the St. Jude Comprehensive Sickle Cell Center Clinic are located on the third floor of the LeBonheur Children's Medical Center Physicians Office Building across from the main hospital. They are staffed by members of St. Jude Children's Research Hospital and the University of Tennessee, Memphis. Administrative offices are located in the Physician's Office Building, Suite 345. The Pediatric Hematology Clinic is located in Suite P110. This Center is an affiliate member of the Sickle Cell Disease Association of America.

Patients: The Pediatric Hematology Division provides comprehensive treatment of infants, children and adolescents with sickle cell disease, as well as other hematological conditions. Services are available to all patients in western Tennessee, northern Mississippi and eastern Arkansas. Almost 800 children and adolescents with sickle cell disease are followed on a regular basis. More than 800 hospitalized patients are treated by the staff annually. Outpatient Pediatric Hematology Clinic visits average more than 1,000 per year.

Research: Clinical research in sickle cell disease is a major component of the Comprehensive Sickle Cell Center. The Center has published more than 60 articles involving both national and local research during the past 20 years.

Education: The Sickle Cell Center has a comprehensive family education program for parents of children with sickle cell disease. This seven-phase program includes home visits, parent-education classes, films and individual teaching sessions. Center activities also include the education of health professionals. Lectures and presentations for nurses, physicians, and counselors are given regularly at local, regional and national conferences. The Center has recently added a full-time school teacher who provides a liaison between the school-age patients with sickle cell disease and their local schools to ensure that the children receive the appropriate educational resources needed.

Genetic Counseling: The Sickle Cell Center offers counseling and education to families of children and to individuals with sickle cell trait. Outreach screening, counseling and education

services are provided to at-risk populations. As part of the Tennessee Department of Health Genetics Program, the Center coordinates sickle cell trait counseling in western Tennessee. The Center has helped establish a network of Health Department nurse counselors who provide trait-counseling services throughout the 21 counties in the western part of the state. Counseling and education are also provided for other hemoglobin trait conditions.

Other Activities: This Sickle Cell Center sponsors an annual summer camp for children with sickle cell disease ages 7-16 years. Campers participate in a wide variety of recreational and educational activities.

Chattanooga Sickle Cell Center at T.C. Thompson Children's Hospital

Director: Manoo Bhakta, MD

The Division of Pediatric Hematology/Oncology at T.C. Thompson Children's Hospital in Chattanooga is an affiliate of St. Jude Children's Research Hospital. The Center provides identification, medical care, client and community education, counseling, follow up of newborn screening and all preventive aspects of care for sickle cell disease and other hemoglobinopathies. The Division provides a resource for all area physicians and facilitates the care of sickle cell patients as well as tracking and counseling of sickle cell trait. This Center provides services for the 11 counties of the southeastern area of the state of Tennessee.

Knoxville Sickle Cell Center:

Director: Carmen B. Lozzio, MD

Sickle Cell Coordinator: Diana White, MSSW

This Center is part of the programs offered by the University of Tennessee Developmental and Genetic Center already described. The Center is providing case management follow up for 10 patients with sickle cell disease and genetic counseling for the families of 70 new infants diagnosed as carriers of sickle cell trait. The Sickle Cell Coordinator provides case management services (home visits, counseling and infants diagnosed with sickle cell trait receive genetic counseling.

THE HEALTH INFORMATION INFRASTRUCTURE:

Children Information Tennessee (CIT) began in 1998, as an integrated data system providing historical and user updated information. CSHCN is the central component of the State Genetics Information Technology Plan. CIT forms a multi-department initiative linking demographic and program participation information from Health, Human Services, Children's Services, Education, and TennCare. Prior to the creation of CIT, there was no single source of information that provided case management professionals with a comprehensive view of program participation by their clients or the ability to communicate directly with each other about their mutual clients. In addition to the state agencies serving children, representatives of other interested government and private entities were members of the CIT Oversight Committee (Commission on Children and Youth, the Center for Effective Government, Family Voices Tennessee, and Tennessee Voices for Children) and participated on the work groups creating the vision and plan for CIT.

CIT currently contains approximately 1.8 million demographic records on children born in Tennessee, in the Immunization registry or in other participating programs. There are currently linked program participation records from TDH (Total 242,860; WIC 51,240, Children's Special Services (CSS) 81,010), Tennessee Department of Children's Services (TDCS) (25,383), TennCare (277,406), TDE (Total 103,973; Early Intervention 6,923, Special Education 97,050), and Human Services (9,207). Access to this information is via secure, encrypted communication channels and controlled by user role and parent/guardian permission and electronic certificates. The central data repository is maintained in a secure computer facility, behind a backside firewall with the web server and authentication server behind a front-side firewall. The central data repository is highly optimized and indexed to provide typical access times to demographic records in less than 10 seconds. For additional information see Attachment A, The TN IT Plan.

PLANNING ACTIVITIES:

The Statewide Genetics Planning Committee (SGPC) was organized to prepare the application for the Tennessee Genetics Planning Grant awarded by the Genetic Service Branch (GSB) of the Division of Services for Children with Special Health Care Needs of HRSA under the "State Development Grants for Newborn Screening Efforts and Infrastructure Development". Tennessee was also awarded a grant for "Data Utilization and Enhancement" by the Office of Data and Information Management of HRSA. In April 2003 TN received a four-year grant for plan implementation that would include integration of the Genetics and IT services. The SGPC is now the Statewide Genetics Coordinating Committee (SGCC).

PROBLEM STATEMENT:

POPULATION:

A description of the population and the challenges related to the frequency and demographic distribution of birth defects and genetic disorders is an important aspect to consider in planning future programs.

The population of Tennessee has increased significantly in the 1990s compared to the 1980s. It was estimated at 5.3 millions in 1995 and 5.7 millions in the year 2000. This increase ranked Tennessee as having the ninth largest in the nation. The projection by the year 2025 is 6.7 million people.

The distribution of Tennessee's population is approximately 17% black, 82% white and 1% other races. However, Tennessee is expected to gain 97,000 people through international migration between 1995 and 2025 and is expected to gain 845,000 people through internal migration for the same time period. All ethnic and racial groups are expected to increase during this time period except for non-Hispanic whites. African-Americans, Asians, and persons of Hispanic origin will experience the greatest gain. According to the 2000 Census, the Hispanic/Latino population in Tennessee was 123,838 representing 2.2% of the state's population. This is twice the number estimated in 1998 by the Census Bureau.

Refugees and legal immigrants are now arriving from African, Baltic, Central Asian and Southeast Asian countries. Southeast Asians are another growing population group (52,564), and the state has the fifth largest Kurdish resettlement site in the nation. Refugees and legal immigrants are now settling in Tennessee from Africa, the Baltic regions, Central and Southeast Asia, Iraq, Somalia, and the Sudan. The state also anticipates an influx of refugees, especially women and children from Afghanistan, Sudan, Burma, and Sierra Leone. These refugees are from war torn countries where rape and torture were commonplace. They are expected to have significant health problems.

Current ethno-cultural barriers include language, educational level, health care customs, and religious restriction against medical intervention. Over 30 different languages are spoken as the primary language of the home in metro-Nashville. Each major medical institution in the state has a network of locally available interpreters, and, statewide, the Tennessee Foreign Language Institute maintains a database of translators and interpreters for a wide range of linguistic needs. Other resources are available in more isolated communities, including the AT&T Language Line, which can be subscribed to (1-800-643-2255). Educational level and health care customs are sometimes barriers. Most medical institutions have found that parents are not resistant to treatment and therapy when the issues are appropriately explained. Religious prohibitions are also rare, but Tennessee law is clear on the obligation of caregivers to bring to the attention of the TDCS any instances in which denied treatment would result in harm to the child.

Tennessee covers 41,220 square miles of land area and is approximately 500 miles from east to west and 110 miles from north to south. The state is divided into 95 counties, each with a health department mandated by state law and located in the county seat. For departmental administrative purposes, the counties are grouped into seven rural and six metropolitan health regions. Topographically, as well as culturally, the state is divided into three grand regions (East, Middle and West).

East Tennessee has 35 counties and contains the Appalachian Mountains at its eastern boundary and the Cumberland Mountains at its boundary to the west. The population of the region is 92% Caucasian, 6% African-American, and 2% of other races.

Middle Tennessee has 39 counties. The eastern counties of Middle Tennessee are part of the plateau of the Cumberland Mountains and the remaining counties are located in a flat fertile rural area. The population is 86% Caucasian, 12% African-American, and 2% all other races.

West Tennessee has 21 counties. The population of this region is made up of 63% Caucasians, 35% African-Americans, and with other races making up the remaining 2%. This area has the largest percentage of sickle cell disease, approximately 75% of the state total.

In summary, the diversity of races, cultures, and the presence of rural and metropolitan areas within each of the three grand divisions of the State represent a challenge and an opportunity to compare strategies that may be useful for the integration of services with a family centered, community based approach. The prevalence and demographic distribution of the majority of birth defects and genetic disorders is unknown. Plans to retrieve some of this information from the data contained in CIT is proposed as part of this project and is one of the goals of the Tennessee Birth Defects Registry.

SPECIFIC AREAS OF CONCERN:

1. Increase in the number of tests screened by NBS programs:

NBS is the largest public health program in the USA since virtually every infant born in the 50 states is screened for at least two diseases. Some states only test for as low as two disorders and others test for as many as thirty. NBS is seeing rapid changes because of the use of TMS, a technique that can test for a significantly larger number of disorders over what has been the standard for many years. These technical advances have created scientific, ethical and economical issues that need to be addressed before recommending that all the state test for as many as 30 additional disorders. The Expert Group of the HRSA funded contract to the American College of Medical Genetics titled "Standardization of Outcomes and Guidelines for NBS Progress" will soon be making recommendations on these issues and states will need to upgrade their programs to meet national expectations.

2. Expansion of the health information system infrastructure:

Prior to the creation of CIT, there was no single source of information that provided case management professionals with a comprehensive view of program participation by their clients or the ability to communicate directly with each other about their mutual clients. A group of state agencies serving children, led by the TDH, were concerned with service fragmentation and duplication. Five participating state entities (Health, Children's Services, Education, Human Services, and TennCare) crafted and signed a Memorandum of Understanding (MOU) creating CIT to address these concerns.

3. Integration of systems of early identification with early intervention:

Several state departments provide services for CSHCN the coordination of these services is a major challenge. The different state departments involved are: 1) TDH including NBS and Genetic Services, CSS, Vital Statistics, Immunization, WIC, and other health related services. 2) TDCS for children in state custody; 3) TDE, including the TEIS; 4) Human Services with the Supplemental Security Income Program; 5) Mental Health and Developmental Disabilities; 6) Finance and Administration which includes the Division of Mental Retardation and the Bureau of TennCare which administers the federal Medicaid funds through a system of managed care organizations (MCO). On November 1, 2000, these departments signed an Interagency Agreement to fulfill the requirements of Part B and C of the Individuals with Disabilities Education Act (IDEA). Interagency agreements have also been signed by the state departments participating in CIT and by those participating in the Statewide Genetics Planning Committee. However, integration of services at the local level for each individual child is still a challenge since different care managers and service providers work with each child and parents are often confused concerning the role of each agency serving their child.

4. Implementation of quality assurance and evaluation of service delivery systems:

Although TDH is already promoting Comprehensive Systems of Services and State Performance Measures are being evaluated for many aspects of the MCH program, specific measures of services for NBS and NHS are needed and will be an important aspect of this proposal.

5. Professional education:

Informing/educating PCPs (medical homes), and other health professionals on NBS and NHS and other genetic disorders is needed to improve services for CSHCN in general and specifically for children identified by NBS and NHS or those who have other genetic disorders. Results of **Focus Groups (see Attachment B)** conducted by Family Voices of Tennessee indicate the need to educate PCPs (medical home) specialists, and other health care professionals.

6. Consumer participation in planning, implementation and evaluation of the genetics and information plans:

The Consumer Involvement Subcommittee collaborating with the Ethics Subcommittee of the Statewide Planning Committee has identified needs and areas of concern to be addressed in this proposal. A **Needs Assessment** conducted in 199 by Family Voices of Tennessee through a survey and the same **focus groups** of 2003 described above (see Attachment B) suggest that one of the barriers to accessing needed services are the limited benefits of both TennCare and commercial insurance plans. The Genetics Subcommittee on Consumer Involvement will be collaborating with Family Voices of TN to conduct surveys of the entire CSS population. Children identified by the NBS and NHS programs as well as children with other genetic disorders will be included. A major challenge faced by Tennessee is a recent ruling by the Bureau of TennCare limiting eligibility of children and adults who are not Medicaid eligible. These children will not be able to have a medical home until CSS and local health departments can restart the direct medical care services they were providing before TennCare began covering medical care for all uninsured children in Tennessee.

GOALS AND OBJECTIVES:

1. Enhance and expand the state newborn screening program:

- a. Tennessee is planning to add four screening tests to the current five NBS and the NHS. The new tests are: Biotinidase, MCAD, Homocystinuria, and MSUD.
- b. Plans for additional screenings for other genetic disorders have been recommended by the GAC and will be implemented after the TMS equipment and software are installed, the personnel is trained and the experience of other states is evaluated by the TDH staff and the Statewide Genetic Planning Committee, and the subcommittees on NBS, Ethics and Consumer Involvement. The national recommendations from the HRSA funded NBS Expert Group will be followed.

2. Expand and enhance the public health information system infrastructure:

- a. Data collected at the state laboratory will be transferred to a central location. This will enhance compatibility, reliability, and integrity of the data. Tennessee plans to improve the linkage process of metabolic and hearing screening records; add hospital patient ID and heal stick card tag numbers to the electronic birth records; modify linkage program to use additional identifiers; and compare linkage quality with and without additional identifiers.
- b. The new HRSA agreement will allow Tennessee to incorporate early identification and early intervention data into the TNCHP based on the expansion of CIT. This includes improving the

linkage process of HDDS; adding hospital patient ID numbers to electronic birth records beginning in 2004; obtaining patient ID numbers, full name and address on patients under six years of age for 2000 through 2003 for linkage to existing HDDS data to birth records; comparing linkage quality with and without supplemental identifiers (2000-2003) and with and without additional identifiers beginning in 2004.

c. Continue to enhance and expand system data availability and applicability. Add EPSDT data, both screening and tracking, to existing child health profile.

d. Improve process for linkage and use of data as required to sustain and improve system. Plans are for the placement of place diagnostic, prescriptions, immunizations, screening, treatment, and follow-up data in structured schema and extract child health profile data from source systems to create time stamped detail records. The use of time stamped detail records will make it possible to create a longitudinal summary at each age point in Tennessee's EPSDT schedule.

e. Add web interface to TNCHP information. This project will make it possible to build a system from the existing data system for child health profiling in a web-based interface. CIT's web site will serve as a portal, controlling access, presentation of information and functionality based on user role and parent guardian permission.

f. Enhance usage and application of data system-wide. Tennessee will add more flexible procedures for obtaining and reporting parent/guardian permission. This requires that the current CIT procedures work satisfactorily in a state entity setting. With the expansion beyond core state and state related entities, (e.g. pediatric practices,), more flexible methods are required. For example, a parent bringing a child in for well baby care could authorize release of child health profile data to the practitioner providing the care.

g. Explore the possibility of making available to the parent/guardian a copy of their child's health profile from the web site. Protocols will be developed to allow parent/guardian (with pertinent permission agreements) to access a copy of their child's information by using a secure PIN number without the involvement of individual case managers. Parents/guardians will be able to access TNCHP information, update permissions and annotate the TNCHP.

h. Automate the identification of CSHCN. Our screening and service programs will identify most CSHCN. Additional children not screened or served may be identified from data sources used by CIT. Data elements, e.g. International Statistical Classifications of Diseases (ICD), Current Procedural Terminology (CPT), etc., will be used to identify CSHCN and children with inheritable conditions. A set of codes and decision rules will be created to identify these children.

3. Integrate systems of early identification with early intervention.

a. Coordination of services at the state department level. The SGCC will incorporate additional members representing government, university and other agencies as appropriate and needed. Inter-agency agreements will be signed and this Committee and subcommittees will develop plans to promote coordination of services.

b. Coordination of NBS with services of early intervention at the local level will be aimed at developing a comprehensive continuum of services from diagnosis and screening to treatment, counseling, coordination with the medical home, case management and Early Intervention and Rehabilitation services.

c. Implementation of the NHS plan, diagnostic assessments and early intervention is an objective of this proposal and the Goal of the Tennessee NHS Plan.

d. Linkage of data on NBS, NHS and genetic services, case management and early intervention to the CIT system. This component of the program will help identify gaps in services and areas of need for better coordination. Once confidentiality issues are resolved the information on services received each child will be available to providers, case managers and parents if they are authorize to access this information.

e. Collaboration with all aspects of the MCH program for CSHCN: This program will be an integral part of all TDH-MCH programs for CSHCN.

4. Implement quality assurance and evaluation of service delivery systems to determine system effectiveness and efficiency.

a. The NBS program reports the number of NBS performed by hospitals along with the percentage of unsatisfactory specimens and reasons why the specimens were unsatisfactory. This report is being revised to include the number of hearing screens performed and the rate of NHS referrals. These data will be incorporated into the CHP and linked to other information available on each infant. This activity will continue to be an integral part of the quality assurance matrix. The integrated data repository holds the demographic, process and outcome data of CSHCN. Standard tools are available for analyses and reporting. These tools will be used to describe the population, track their progress through the system, identify gaps in services and monitor outcomes. The project will also create standard reports describing person, place, and time of genetic services within the system and publish aggregate reports on the web.

b. The hospitals with Universal NHS programs will be monitored and the data collected and examined for completeness and accuracy.

c. Services initiated by TEIS for children identified by hearing and metabolic screening will be tracked.

d. The project staff will create a research agenda by evaluating the characteristics of the population at risk and tracking of service needs deemed appropriate. Incidence of occurrence will also be studied and plans formulated to ensure increased health care provision for CSHCN.

5. Increase provider's understanding of the importance of timely and appropriate treatment for children with genetic conditions.

a. Inform health-care professionals about current newborn screening program requirements, the immediate responses required by Genetic/Metabolic or Sickle Cell Centers, endocrinologists and primary care professionals; inform of resources available, and the Center's medical home long-term follow-up; interaction of positive newborn screened infants.

b. Educate Health Care Professionals about general genetic services in TN including set up additional Genetic web-based information on the TN-MCH web site with links to NBS information.

- c. Provide relevant culturally sensitive information on Genetics and genetic services in TN to professionals and families

6. Empower families to be effective advocates for appropriate care and services for their CSHCN.

- a. Enhance parental access to and control over information.
- b. Ensure that parents know about newborn screening and about their rights to information about their child's care.
- c. Improve cultural competence of care for children with special health care needs (CSHCN).
 - a. Increase awareness of resources, providers, and support groups for families and affected children, adolescents and adults.
 - b. Inform consumers of anticipated additions to the Tennessee Newborn Screening Test Panel
 - c. Decrease the percent of families of CSHCN that report problems and/or are dissatisfied with services.
 - d. Increase the percentage of CSHCN that has seen their PCP at least once.
 - e. Increase utilization of early intervention services.

METHODOLOGY:

The implementation of the Genetics Plan and the Information Technology Plan with integration of both plans to achieve the goals and objectives of this proposal will be performed as follows:

1. Strengthen and Expand the NBS Program

a. Add four genetic tests to NBS: Plans to implement the expansion of the NBS program to add four new tests are already in progress and will be implemented during the year 2003. The specific activities will be carried out by the personnel of the TDH, NBS Laboratory. Two different types of equipment, and methodologies are required to screen for Biotinidase and for three metabolic disorders (homocystinuria, MSUD and MCAD) to be tested by TMS. Bids for the purchase of the two types of equipment have already been requested. Follow-up with the Purchasing Department to award the contracts and with the vendors to install the equipment and software will be done in the next two to four months. Training of the staff, set up of pilot studies, validation of results and incorporation of the new tests to the forms used for collection of the blood specimens will be carried out during 2003. Drafts of the literature to be distributed to parents including the new disorders have already been prepared and reviewed by the Genetic Advisory Committee. This planned expansion of the NBS program to include a total of nine (9) genetic tests and hearing screening will place Tennessee among the states in compliance with the MOD recommendations for minimum standards for NBS programs.

b. Include additional tests to NBS: Additional tests will be added after the four new tests are implemented. Expansion to include additional tests has been recommended by the GAC and will be based on the use of TMS and on scientific, ethical, and appropriate health outcomes for individuals, families and society rather than technical feasibility of performing the tests. We plan to follow the national recommendations to be developed by the NBS Expert Group of the HRSA funded contract to the American College of Medical Genetics (ACMG) entitled “Standardization of Outcomes and Guidelines for State Newborn Screening Programs”. The general goals of this Expert Group are similar to those discussed in our State by the Ethics and Consumer Subcommittees of the Statewide Genetics Planning Committee and the Genetics Advisory Committee

2. Expand and enhance the public health information system infrastructure.

TDH IT System is called CIT and the operation of this system is described as follows: While the creation of CIT predates the National Electronic Disease Surveillance System (NEDSS), it is very similar in philosophy and implementation (see IT Plan: Attachment A) Confidentiality is a very important aspect of this system and the Ethics/Confidentiality subcommittee has made recommendation and reviewed the impact of the Health Insurance Portability and Accountability Act (HIPAA). Our current IT system (CIT) security, privacy and tracking of access to information already meet the current guidelines. We have executed Memorandums of Understanding (MOU) with all agencies/departments that participates in CIT. These MOUs give us the legal authority to share data on individuals who are served by the participating agencies. Thus, information collected by the participating agencies/programs may be stored in CIT. Further, we have added parental/guardian permission to the system to control access to the information stored in CIT. Individuals within an agency/department have the same level of access to CIT stored information as they would in accesses their agency/department's information system. Parental/Guardian permission controls the cross entity access. For example, if permission has been given to all entities other than Children's Services to share data, a Children's Services case manager would not see any not Children's Services program participation information.

See the TN IT Plan, Attachment A, for additional information

3. Integrate Systems of early identification with early intervention

The integration of early identification and diagnosis of disorders screened by the NBS program to all other MCH services provided to CSHCN and early intervention services will be carried out as follows:

a. Coordination at the State Departments Level:

The Statewide Genetics Planning Committee will become the **Statewide Genetics Coordinating Committee**. (SGCC) This multidisciplinary committee, advises state officials and is involved in the review of new tests and the ongoing evaluation of all aspects of the state's process for newborn screening and evaluation and monitoring of activities. This committee has 29 members and includes 12 programs within the TDH (MCH, CSS, Genetics and NBS, Birth Defects Registry, Vital Records, Immunization Program, Cancer Registry, Community Education, and BHJ, Representatives from the Genetics Advisory Committee, and the CSS Advisory Committee and from the TDE, Department of Mental Health and Developmental Disabilities, and the Division of Mental Retardation are members

of this committee. Consumers organizations represented include the Alliance of Genetic Support Groups, TN Voices for Children, Family Voices of TN, and March of Dimes. Additional members representing other state departments such as the TDCS and Bureau of TennCare will be invited to participate and to sign interagency agreements. State legislators with interest in NBS, NHS and genetic services will also be invited to become members of this committee.

Most of the activities of this committee are performed by subcommittees. An Executive Committee that includes the three co-directors of this program, the program coordinator, and the chair of the subcommittees will be actively involved in the monitoring and evaluation of all aspects of this program.

b. Coordination of NBS with services of early intervention at the local level:

The following process for follow up of NBS results combines current practices and proposed improvements:

- 1) For all presumptive positive results the state staff notifies immediately by phone and fax the PCP and either the Genetic/Metabolic Center, the Sickle Cell Center, or the Pediatric Endocrinologist depending on the type of disorder.
- 2) The staff from the centers and the PCP notifies the families and requests a repeat specimen or obtains a sample for confirmatory diagnosis depending on the established protocols for each disorder. Critical values for disorders such as classical galactosemia are considered a medical emergency and the families are notified immediately and treatment of the infant is started before results of the confirmatory tests are completed.
- 3) Cases with a confirmed diagnosis are treated at the respective regional centers for metabolic disorders and sickle cell or other hemoglobinopathies. The regional centers have medical genetic specialists and metabolic nutritionists for treatment of metabolic disorders or pediatric hematologists for treatment of sickle cell and other hemoglobinopathies. Pediatric endocrinologists act as consultants to PCPs and the treatment for hypothyroidism and congenital adrenal hyperplasia is monitored by the PCPs with consultation as needed from the pediatric endocrinologists.
- 4) All infants with a confirmed diagnosis are followed by the PCPs (medical home) for pediatric care and the specialty care is managed by the specialists at the regional centers.
- 5) The regional centers have genetic counselors and genetic social workers who provide information about the specific disorder and referral to Family-to-Family support groups and to case managers in other programs such as CSS, TEIS, and Family Voices of TN.
- 6) Web access to information on NBS, NHS and genetic disorders will be available at the TDH MCH, Genetics and NBS web page. This website will be updated and detailed information on proposed additions is described in the reports of the Professional and Consumer Involvement subcommittees.
- 7) The data on services provided will be entered in the Child Health Profile and will be used to facilitate coordination of follow-up services by providing case managers, parent/guardians and service system managers with a more comprehensive picture of children's needs and the services they are receiving. The TNCHP will add both breadth and depth to what is currently collected and retrievable on individual children. Improved analysis, reporting and visualization of the TNCHPs and supporting data will help institutionalize, promote and disseminate the formative and summarized evaluation of system and individual service delivery performance. The goal is to provide appropriate

and timely access to information needed to improve service delivery, emphasizing both strengths and weaknesses.

8) Providers with approved access to the data will be able to retrieve authorized data at the point of service through standard reports and system queries.

9) Parents/guardians will be able to have secure and confidential access to their child's health profile.

The above description of services and the service flow chart combines current procedures for diagnosis and treatment of genetics disorders detected by the NBS program and proposed referrals for case management and early intervention services as well as the role of the data information plan to assure a continuum of service delivery for families with newborns or children with or at risk for genetic disorders.

Confidentiality is a very important aspect of this system and the Ethics/Confidentiality subcommittee has made recommendation and reviewed the impact of the Health Insurance Portability and Accountability Act (HIPAA). Our current IT system (CIT) security, privacy and tracking of access to information already meet the current guidelines. We have executed Memorandums of Understanding (MOU) with all agencies/departments that participates in CIT. The MOUs give us the legal authority to share data on individuals who are served by the participating agencies. Thus, information collected by the participating agencies/programs may be stored in CIT. Further, we have added parental/guardian permission to the system to control access to the information stored in CIT. Individuals within an agency/department have the same level of access to CIT stored information as they would in accesses their agency/department's information system Parental/Guardian permission controls the cross entity access. For example, if permission has been given to all entities other than Children's Services to share data, a Children's Services case manager would not see any not Children's Services program participation information.

c. Implementation of a statewide system for NHS:

The NHS Program has a plan similar to the NBS for integration of screening, diagnoses, and follow-up services. The differences are the following: Confirmatory diagnosis is performed by audiologists and ENT specialists and referral to the genetic center is made after confirmation of the diagnosis in order to determine if a genetic disorder is the cause of the hearing loss. In this case, the geneticists are consultants and the treatment is the responsibility of the ENT specialists, audiologists, and PCPs. A plan for expansion of the NHS program is enclosed in the attached Work Plan Matrix.

d. Linkage of data and development of the Child Health Profile

The federation of data sources covered by CIT provides the basic building blocks to create a child health profile. The child health profile is the organizing principle used to bring together demographic, diagnosis, treatment and outcome data on CSHCN. The child health profile will be created over time and permit a longitudinal look at the developmental process for CSHCN. The child health profile will have data for tracking children over time, assessing the effectiveness and efficiency of the service delivery system, and measuring short and long-term outcomes. (See Attachment B-IT Plan)

e. Collaboration with all aspects of the MCH program for CSHCN:

The newly funded HRSA grant will be an integral component of the state's MCH program for CSHCN. A strong coordination and collaboration already exists between the Genetics Program and

the state's IT program which includes the NEDSS initiative. Collaboration and a working relationship will also be established with the CDC Bio-terrorism (BT) Coordinators and local BT and NEDSS committees. The Child Health Profile will integrate data on immunizations, registries, vital records, birth defects, chronic diseases, early identification, WIC, TennCare (state Medicaid), and other related programs. To automate the identification of CSHCN who have heritable conditions, current classification schemes using ICD codes will be assessed and modified to work with Tennessee's population. This will make possible a linkage of diagnosis to services as well as to determine demographic distribution and prevalence of genetic disorders.

4. Implement quality assurance and evaluation of services delivery systems to determine system effectiveness and efficiency

A detailed Work Plan Matrix has been created for each goal and will be used by the staff and the members of the SGCC committee and subcommittees to monitor and assess the achievement of these tasks. The State Assessment Tool produced by the Council of Regional Networks for Genetic Services (CORN) (<http://www.cc.emory.edu/PEDIATRICS/corn/new/guidelns.pdf>) will be also used to compare the state genetic services to those outlined in the CORN Guidelines for Clinical Genetic Services for the Public's Health.

Outcomes for the target population will be measured and improvements in service delivery systems and in effectiveness and efficiency will be based on achievement of the proposed health outcomes. Reductions of morbidity and mortality and increase in the number of services provided, in the number of disorders screened and in the coordination of services will be monitored and evaluated.

Some of the health outcomes that this Plan aims to achieve are:

- Increase in the number of infants diagnosed and treated with various metabolic disorders detected by expanded screening using TMS
- Assurance that all infants who have an abnormal NBS or NHS are followed, the diagnosis is confirmed and they are treated and receive coordinated care.
- Integration of NBS, NHS and other genetic services into health promotion programs and preventive care services
- Education of health care professionals and consumers through printed materials, web-based sources, lectures and media announcements.

Examples of the type of the type of research that the staff will be investigating are:

- Determination of the frequency of specific metabolic disorders in each region of the state (east, middle, west) to detect clustering and to investigate possible causes.
- Assessment of the incidence of congenital hearing loss in each region of the state and the percentage of hearing loss caused by genetic disorders.

- Evaluation of the incidence of other genetic disorders identified by the Birth Defects Registry and by the Genetic Centers, to identify clusters of disorders and to investigate probable etiologic factors.
- Development of an automated identification of genetic diagnoses and CSHCN based on ICD-9 diagnostic codes to track information on service utilization, case management, participation in early intervention services and insurance coverage.

Analysis, visualization, and reporting of data will continue to be an integral part of the quality assurance matrix. The results will be readily available to key stakeholders. The staff will create standard reports describing person, place, and time of genetic services within the system and publish aggregate reports on the web.

5. Increase provider’s understanding of the importance of timely and appropriate treatment of children with genetic conditions.

- Revise the existing NBS pamphlet describing the five disorders currently screened in Tennessee and includes the additional disorders to be screened. Provide opportunity to health care professionals for feedback on brochures and periodic review of comments by NBS personnel and GAC when needed.
- Update and continue distribution of the video “Let’s do it right the first time” to hospitals for viewing by technical and nursing personnel, with additional print inserts about the new tests to be offered and revise this tape to include the new disorders.
- Consider adaptation of medical information in above tape for medical professionals education and explore the use of CDC NBS/educational site or link and video for educational programs to local medical societies, hospitals and health departments.
- Develop Information sheets on each of the disorders screened using medical information in the educational tape as well as the American Academy of Pediatrics (AAP) Summaries. Develop and prepare Emergency Fact sheets for the four additional disorders to be screened.
- Include information on NHS and NBS on all brochures and in the educational video that emphasizes the need for follow up evaluation of a positive hearing screening and the genetic implications of a diagnosis of congenital hearing loss.
- Make available all information sheets in print and web-based sources to be distributed to PCPs, Perinatal Centers, Public Health Departments, TennCare MCO Directors, and other relevant individuals in their organizations, hospital laboratories, hospital nurseries/birthing units; Tennessee Medical Society, Tennessee Nursing Organization, educational institutions including the departments of Pediatrics, Family Practice, and Internal Medicine, Nursing Schools, Med Tech Schools, Medical Centers etc. for use in educational endeavors.
- Place all finalized sheets/forms/information and written brochures on the existing TDH, NBS website, with counters for tracking 'hits'. Make all NBS forms currently faxed available on the TDH, NBS web site. Add information about and links to Regional Genetic Centers and Sickle Cell Centers. Develop links to appropriate information about NBS and further information about genetic disorders (see Appendix: Professional Education Subcommittee Report and Web Links for Professionals).

Include the links for Consumers (see Appendix on Consumer Involvement Subcommittee and Web Links for Consumers). Make available to users a “Comments” text box for them to provide feedback on forms/information and links.

h. Publish information in available resources about Newborn Screening initiatives. For example the TDH Newsletters, Tennessee Medical Association (TMA) Journals and TN Nursing Publications/Newsletters

i. Provide direct contact information by TDH phone/fax and face-to-face didactic talks by GAC members/Genetic Centers at workshops, lectures, classes, one-on-one, - Medical/Nursing society/meetings - Use of "Let's Do It Right the First Time" video, educational portion only.

j. Distribute information to Media (Newspaper releases- Media interviews) as approved by GAC/NBS program.

6. Empower families to be effective advocates for appropriate care for their CSHCN

a. Develop or coordinate development of a brochure with web sites, support groups, etc.; translators will be utilized as necessary; submit materials to the TDH for approval prior to distribution and/or printing.

b. Members of the Genetics Consumer Involvement Subcommittee will assist the TDH as needed to incorporate additions to newborn screening panel on brochures, web sites, educational presentations, etc.

c. Disseminate information in English and Spanish to consumers through Genetic Outreach Clinics and consumer organizations.

d. Translators will be engaged to develop materials in additional languages as deemed necessary.

e. Conduct focus groups in each genetics region with families who have used specialty care, mental health services, physical therapy, occupational therapy, speech therapy and home health services to define the problems encountered in securing services, make recommendations to fix the problems, and develop a report of findings with recommendations for improvement – and – use results of focus groups being coordinated by the Family Voices Family to Family Grant to further define problems and develop strategies to improve satisfaction.

f. Present results of focus groups and submit to professional newsletters for publication. Staff of Genetics Centers and members of the Statewide Genetics Committee will serve as speakers for presentations to constituent advocacy groups and specialty services associations. Participation in presentations will be documented.

g. Members of the Genetics Consumer Involvement Subcommittee will develop or coordinate the development of a PowerPoint Presentation and brochure; translators will be utilized as necessary; and materials will be submitted to the TDH for approval prior to distribution and/or printing.

h. Distribute a report of focus group findings and suggestions for empowering families in seeking satisfaction in how their health care needs are addressed.

- i. Post list of resources for families of CSHCN on the TDH web site.
- j. There will be continuing interaction with the Family Voices Family-to-Family grant (FVFFG) and Tennessee Genetics Consumer Involvement Subcommittee via shared liaison(s). Report of focus groups (see appendix for locations) will be available through shared liaison(s) to avoid duplication of services.
- k. Members of the Genetics Consumer Involvement Subcommittee will serve as liaison with TEIS for purposes of coordinating activities and as liaison with CSS and school-based intervention services directors.

PLAN DEVELOPMENT:

THE STATEWIDE GENETICS PLANNING COMMITTEE (SGPC)

The Statewide Genetics Planning Committee has 29 members and was established in the year 2000 with the charge to develop a comprehensive genetics plan. See Attachment B for members and their affiliation. It includes representatives from 12 programs within the Department of Health (Maternal and Child Health, Children's Special Services [CSS], Genetics and Newborn Screening, Birth Defects Registry, Vital Records, Immunization Program, Cancer Registry, Community Education, and Bureau of Health Informatics). Representatives from the Genetics Advisory Committee, and the CSS Advisory Committee and from the Department of Education, Department of Mental Health and Developmental Disabilities and the Division of Mental Retardation are members of this Committee. Consumers organizations represented include the Alliance of Genetic Support Groups and TN Voices for Children. This committee has been expanded and subcommittees have been established to address specific aspects of the plan.

SUBCOMMITTEE STRUCTURE

Newborn Screening

This subcommittee is composed of all eleven members of the Genetics Advisory Committee (GAC). It is made up of all the directors of the genetic and sickle cell centers, two at-large members and others including consumers and representatives of consumer advocate organizations, who receive a standing invitation to meetings and teleconferences. This sub-committee, because of its established link with the commissioner, will have oversight responsibility for the work of all other subcommittees. Their role will also include recommendations and priorities for adding additional screening tests. Chaired by Mitzi Lamberth, RN, Director of Genetics and Newborn Screening, MCH, TDH.

Financing Newborn Screening Services

Expansion of newborn screening services cannot occur without additional funding. Currently the State Laboratory charges hospitals \$ 17.50 for each initial screen. This fee will be raised when the four additional tests are implemented. The fee is based on cost for the performance of the laboratory tests and the tracking and follow-up of infants with presumptive positive results. Chaired by Jim Gibson, Director of Microbiology, Laboratory Services, TDH.

Data Collection/Child Health Profile

A key component for improving and integrating services is the ability to track the children (and their families) who are identified with confirmed conditions to ensure that their care is comprehensive and timely. This integrated data system is designed to allow providers, with permission from parent or guardian, to have access to client service data. The Bureau of Health Informatics is responsible for developing and implementing this data system called Children's Information Tennessee, (CIT). Chaired by Richard Urbano, PhD, Assistant Commissioner of the Bureau of Health Informatics.

Ethics/Confidentiality

Numerous ethical and confidentiality issues must be considered and resolved based on requirements by federal and state law, as well as, protection of privacy for the children and their families. Chaired by Ellen Clayton, MD, JD, Professor of Law and Pediatrics and Director, Center for Genetics and Health Policy, Vanderbilt University and member of GAC.

Professional Education

Focus groups conducted with families, who have a child with a genetic condition, identified the need for education of all professionals about genetic conditions. This sub-committee will develop plans of action for educating various professional medical groups about genetic conditions. Target groups are primary care physicians, family practice physicians, pediatricians, nurses, midwives, third party payors, MCOs, and HMOs. Chaired by Jewell Ward, MD, PhD, Director, Division of Medical Genetics, UT Memphis and member of GAC.

Hearing Screening

This subcommittee is composed of the already established Newborn Hearing Task Force (15 members). There are representatives from genetics, pediatrics, neonatology, nursing, audiology, and parents of children with hearing loss, early intervention specialists for the deaf/blind and speech/language pathologists. Dr. Carmen Lozzio represents the state genetics program. Dr. Mark Gaylord represents the Tennessee Chapter of the Academy of Pediatrics and Dr. Brian Carter represents the NHS for this organization. Chaired by Jacque Cundall, RN, Coordinator, Newborn Hearing Screening, MCH, TDH.

Consumer Involvement

Consumer involvement and input in the planning process is critical to meeting the needs of the families and children with genetic conditions. Other individuals and groups needing of education about children with genetic conditions include nurses, special educators and other school personnel, care coordinators, medical social workers. Co-Chaired by Pearl Hann, Director of Program Services, March of Dimes, TN Chapter; Cheryl Major, RNC, BSN, Neonatal Outreach Coordinator, Vanderbilt Children's Hospital and President of the Tennessee Perinatal Association, and, John Phillips III, MD, Director, Division of Medical Genetics, Vanderbilt University and member of GAC.

The subcommittees have conducted business via numerous conference calls and periodic meetings. The reports for some of these subcommittees are attached.

SUBCOMMITTEE REPORTS:

NEWBORN SCREEING SUBCOMMITTEE

MISSION STATEMENT:

- To enhance, expand and improve the State Newborn Screening (NBS) program by integrating NBS and Newborn hearing Screening (NHS) with all aspects of Maternal and Child Health Services (MCH) for children with special health care needs (CSHCN) and with other government, university and agency programs and services.

PROBLEM:

- NBS is the largest public health program in the USA since virtually every infant born in the 50 states is screened for at least two diseases. NBS is seeing rapid changes because of the use of Tandem Mass Spectrometry (TMS), a technique that can test for a significantly larger number of disorders over what has been the standard for many years. These technical advances have created scientific, ethical and economical issues that need to be addressed. The Expert Group of the HRSA funded contract to the American College of Medical Genetics titled "Standardization of Outcomes and Guidelines for NBS Progress" will soon be making recommendations on these issues and states will need to upgrade their programs to meet national expectations.

GOAL:

- Enhance and expand the State Newborn Screening Program

OBJECTIVES:

- Tennessee is planning to add four screening tests to the current five NBS and NHS. The new tests are: Biotinidase, MCAD, Homocystinuria, and MSUD.
- The Genetic Advisory Committee (GAC) at the May 8, 2003 meeting has recommended additional screenings for other genetic disorders detected by TMS.

METHODOLOGY:

- To add four genetic tests to NBS: Plans to implement the expansion of the NBS program to add four new tests are already in progress and will be implemented during the year 2003. The personnel of the TDH, NBS Laboratory, will carry out the specific activities. Two different types

of equipment, and methodologies are required. The equipment to test for Biotinidase has been purchased and training of personnel and validation of results are currently in progress. Testing for this disorder will start by July 1, 2003. Bids for the purchase of TMS are in progress. Training of the staff, set up of pilot studies, validation of results and incorporation of the new tests to the forms used for collection of blood specimens will be carried out before the end of 2003. Drafts of the literature to be distributed to parents including the new disorders have already been prepared and reviewed by the GAC. This planned expansion of the NBS program to include a total of nine (9) genetic tests and hearing screening will place Tennessee among the states in compliance with the March of Dimes recommendations for minimum standards for NBS programs.

- Future expansion of the NBS program to add additional tests will be based on the use of TMS and once this equipment is installed, personnel are trained, and additional software is installed, testing will begin for other disorders. The experience of other states such as California and Massachusetts will be reviewed and national recommendations from the HRSA funded NBS Expert Group will be followed.

COORDINATION:

- The NBS subcommittee coordinates activities with all the other subcommittees of the Statewide Genetics Coordinating Committee (SGCC). A coordinated system of NBS follow-up and treatment has been established as follows:
 - 1) For all presumptive positive results the state staff notifies immediately by phone and fax the PCP and either the Genetic/Metabolic Center, the Sickle Cell Center, or the Pediatric Endocrinologist depending on the type of disorder.
 - 2) The staff from the centers and the PCP notifies the families and requests a repeat specimen or obtains a sample for confirmatory diagnosis depending on the established protocols for each disorder. Critical values for disorders such as classical galactosemia are considered a medical emergency and the families are notified immediately and treatment of the infant is started before results of the confirmatory tests are completed.
 - 3) Cases with a confirmed diagnosis are treated at the respective regional centers. The regional Genetic/Metabolic Centers have medical genetic specialists and metabolic nutritionists for treatment of metabolic disorders. The Sickle Cell Centers have pediatric hematologists for treatment of sickle cell and other hemoglobinopathies. Pediatric endocrinologists act as consultants to PCPs for the treatment for hypothyroidism and congenital adrenal hyperplasia.
 - 4) All infants with a confirmed diagnosis are followed by the PCPs (medical home) for pediatric care and the specialists at the regional centers manage the specialty care.
 - 5) The regional centers have genetic counselors and genetic social workers who provide information about the specific disorder and referral to family to family support groups and to case managers in other programs such as CSS, TEIS, and Family Voices of TN.
 - 6) Web access to information on NBS, NHS and genetic disorders will be available at the TDH MCH, Genetics and NBS web page. This website will be updated and detailed information on

proposed additions are described in the reports of the Professional and Consumer Involvement subcommittees.

EVALUATION AND MONITORING:

- The Co-director for Genetic Services and TDH-NBS staff will monitor the progress for the implementation of testing for four additional disorders. The progress made will be reported to the SGCC Executive Committee monthly and to the SGCC and GAC at their meetings in May and November.
- The NBS, Ethics, and Consumer Involvement subcommittees will evaluate the national recommendations of the HRSA-ACMG Expert Group and will report to the SGCC and GAC. The cost involved in testing and follow-up for additional disorders will be considered and recommendations will be made to the Commissioner of the TDH for implementation.
- Monitoring and evaluation of results and of the FU information on all the disorders tested by TN-NBS program will be performed by TDH-NBS staff and will be presented to the SGCC and GAC at the Bi-annual meetings of these committees.

DATA COLLECTION AND CHILD HEALTH PROFILE SUBCOMMITTEE

See Information Technology Plan (Attachment A)

ETHICS/CONFIDENTIALITY SUBCOMMITTEE

MISSION STATEMENT:

- To ensure that the concerns of children and their families are appropriately addressed in the state newborn screening program

PROBLEM:

- Despite the value of centralized data collections in promoting the appropriate care and follow up of children, parents may be unwilling to permit their child's data to be put in a system such as CIT unless they can be assured that this data will be accessed only by those who need it and used only for the benefit of children

GOALS:

- Ensure that parents know about the care their child is receiving and can be confident that information about this care is used appropriately.
- Ensure the efficacy of procedures for data security and protection of confidentiality

- Assure that children receive culturally competent care

OBJECTIVES:

- Ensure that parents know about newborn screening and about their rights to information about their child's care
- Enhance parental access to and control over information
- Ensure that the CIT achieves its promised protections of data security and confidentiality
- To improve cultural competence of children with special health care needs.

METHODOLOGY:

- In consultation with consumer groups, develop strategies to educate parents about newborn screening, follow up care, and accessing information about their child's care.
- In collaboration with CIT development team, formalize criteria for data security and protection of confidentiality and develop strategies to assess the efficacy of these procedures
- Develop curricula, based on work that has already been done for genetic counselors, to educate clinicians of all types about culturally competent care.

COORDINATION:

- Much of this work will be done in conjunction with consumer groups, such as Family Voices, as well as with the CIT and local physician groups.

EVALUATION AND MONITORING:

- Since the focus is on protecting the interests of families, the assessments will consist primarily of periodic surveys of parents to assess their understanding of the care their child is receiving and to learn about any unmet concerns about care and/or data management.
- Assess the frequency with which parents access the state database.
- Assess the data security and confidentiality of the CIT

PROFESSIONAL EDUCATION SUBCOMMITTEE

MISSION STATEMENT

- Education of health-care professionals in Tennessee about clinical and educational resources of: newborn screening (current/planned), General Genetic Centers/services, and primary care provider partnering with Genetic/Endocrine/Hematology Centers.

PROBLEMS

- *Diverse group of health-care and other professionals* to educate, including: physicians (primary care, Perinatal Centers, specialties, public health clinics); nurses (public health, perinatal units, nurse practitioners); paramedical (nutritionists, social workers, PT/OT, speech therapists, psychologists); teachers (preschool, primary, secondary); third-party payers (private insurance, TennCare Medical Directors, etc.); and legislators.
- *Multiple outlets of information distribution required*, such as: print-based (pamphlets/direct mailings/faxes), TDH brochures/newsletters, web-based, A-V based (videotapes), direct phone/fax contact; direct face-to-face didactic lectures/talks, medical literature (TMA Journal, Tenn. Nursing), and media exposure (newspaper articles/TV interviews).
- *Diverse type information required* for wide variety of professionals.
- *Speed at which the information is needed varies among the disorders*, from emergency (24/7/365) to only needing for occasional reference.
- *Content of scientifically based educational material* can change rapidly, requiring ability to communicate rapidly to a wide variety of professionals.

GOALS

- To educate the health care or public professional about the need to identify disorders early (e.g. Newborn screening), as well as other genetic disorders and birth defects, effects which may be alterable by: early identification, early intervention, and use of available resources (genetic services, public health resources, consumer resources).

OBJECTIVES

- *Inform health-care professionals* about 1) current Newborn Screening Program requirements (responses by the Genetic/Endocrine/Hematology Centers and the Primary Care Professionals, resources available, and Centers' and medical home long-term follow-up and interaction with presumed positive newborn screened infants); and 2) general genetic services at the 5 Genetic Centers.
- *Revise/develop multiple information resources* for above, such as, print, web-based, audio-visual, direct contact, and commercial media.
Participate in outcome assessment, with tracking: the number of products printed/mailed/faxed, hit counter at TDH web-site, the number of individuals trained by Centers, and other outcomes as requested by the Planning Grant.

METHODOLOGY

- Revision of existing NBS information by NBS personnel, and GAC): revise 1) current NBS professional document; 2) relevant information on state NBS Rules and Regulations; 3) institution/hospital responsibilities; 4) the State Lab NBS Follow-up responsibilities; 5) primary Care Providers responsibilities; 6) Perinatal Center responsibilities, 7) the Referral Center information for Genetics/Endocrine/Sickle Cell Centers, and revision of parent NBS pamphlet (NBS personnel, GAC, consumer group).
- Continue dispensing "Let's Do It Right the First Time" video to hospitals for technical and nursing personnel, with revised inserts about new tests. Revision of video to include the additional screened disorders and new information needed to ensure appropriate acquisition of specimens (e.g. transfusion issues). Consider adaptation of medical information in above video for medical professionals.
- Revision of NBS Program presumed instructions (computer generated or faxed info) to PCP/PHD/Perinatal Center for presumed follow-up specimens (TDH, GAC).
- Development of Fact Sheets on existing (revision) and genetic disorders being added (GAC/Metabolic Centers), with uniformity to medical information in above video and AAP Summaries, to contain: 1) relevant medical information; 2) protocol for presumed positive (by center, by PCP/Perinatal Center); 3) long-term follow-up protocol (by center, by PCP/Perinatal Center); 4) Metabolic Center responsibilities; and 5) Medical Home responsibilities.
- Develop additional Emergency Fact sheets for new Genetic disorders to be screened for immediate protocol handling information (Metabolic Centers), and revise existing Emergency Handling sheets as necessary (GAC)
- Hearing Screening education, to include: 1) linkage to existing information on TDH for PCP/Perinatal Centers; 2) provide information on acquiring print information on Hearing Screen; 3) provide education targeted to audiologists; and 4) reinforce available information about evaluation of positive hearing screen, including indications for Genetic evaluations.
- Make available all information sheets in print and web-based sources, specifically to: 1) mail-out information to primary care providers, Perinatal Centers, Public Health Departments, TennCare MCO Directors and other relevant individuals in their organizations), Hospital Laboratories, Hospital Nurseries/Birthing Units, Tennessee Medical Association, Tennessee Nursing Organization, and others, such as appropriate legislators; 2) information packets to be sent to educational institutions (all Genetic Centers, Endocrine, Hematology, General Pediatric, Family Practice, Medicine, Pediatrics, Nursing Schools, Med Tech Schools, Medical Centers etc for use in educational endeavors (list developed by GAC/NBS Program); 3) place all finalized sheets/forms/information on TDH, NBS web-site, with counters for tracking 'hits' (MCH/NBS Program), including Fact Sheets, forms, documents, brochures, and regional Genetic/Endocrine/Hematology Centers contact information/links; 4)develop links to national NBS information and information about specific disorders; 5)explore use of Center for Disease Control NBS CD/educational site or link; 6) develop links to appropriate information sites for general genetic disorders (MCH, GAC); 7) provide links to Consumer Education group pages and Genetics Alliance of Support Groups for provision of Consumer Information (GAC/MCH); and 8) make available to users 'Comments' reply to feedback on forms/information (GAC/NBS program)

- Publish information in available resources about Newborn Screening initiatives: TDH Newsletters (TDH/NBS Program), TMA Journal (GAC/Metabolic Centers), TN Nursing Publications/Newsletters (GAC/Metabolic Centers/Nursing Input), as examples.
- Direct contact information to be provided by: TDH phone/fax, and face-to-face didactic talks by GAC members and Genetic Centers (educational venues, such as workshops, lectures, classes, one-on-one, and talks given to Medical/Nursing societies/meetings). Use of revised "Lets Do It Right the First Time" video. Media distribution as approved by GAC/NBS program, to include newspaper releases, and media interviews. Develop appropriate training to Perinatal Centers and initiate tool for their educating perinatal nursing, and in turn their education of local PCP and PHD.
- Periodic review will be performed by GAC of educational material, both print and web.

COORDINATION

- Professional education works closely with Consumer education to establish information on web sites, coordinate links, and enable medical home to access information for consumers. GAC and TDH to together revise information on NBS information. GAC, MCH and NBS program work jointly to finalize content and implement all information resources. Perinatal Centers informed of educational endeavors and in-put solicited.

EVALUATION AND MONITORING

- Printed Material: TDH-NBS enumerates *printed materials* on newborn screening (to hospitals, birthing units, pediatric hospitals, primary care offices, academic teaching centers, public health departments, and others); review of mailings to additional professionals will be reviewed. TDH will log number and content of *faxes* sent to professionals. Number of *calls* that TDH currently logs about Newborn Screening to Centers' staff, PCP, and parent will be documented. *Other* material, such as the video created by the state, revised video, and information on the other resources, such as web-based materials will be tracked.
- Web-based: a counter should be set up on the NBS web site for enumerating the number of 'hits' (accessions) encountered on the site for specific dates.
- Direct: The Centers provide education enumeration in quarterly reports to the state.
- Media (approved): *Print* media announcement of additional services (e.g. newborn screening initiatives) may be published in a variety of professional and/or general media outlets. The circulation of these vehicles can be used for number of individuals reached. These could include, but not exclusive to: newspaper, newsletter, professional journals, and web-based sites. *Audio-visual media*, such as TV/radio announcements/interviews, would also have estimated target audiences, for number people reached.
- GAC will welcome and review periodically Health Professional and Consumer feedback, from and about any vehicle of educational material.

Web Links – Professional
Tennessee Department of Health website:
<http://www2.state.tn.us/health/MCH/genetics.htm>

NEWBORN HEARING SCREENING SUBCOMMITTEE

MISSION STATEMENT:

- Implement a sustainable statewide Universal Newborn Hearing Screening Program for the early detection and intervention of hearing loss.
- Increase to 95% the number of Tennessee infants that have access to universal hearing screening prior to one month of age.

PROBLEM:

- No active surveillance of newborn hearing screening in Tennessee prior to 2001. No state mandate to conduct newborn hearing screening.

GOALS:

- Implement a statewide system for newborn hearing screening, diagnostic assessment and early intervention.

OBJECTIVES:

1. Develop standards and protocols for newborn screening, diagnostic assessment and intervention.
 - The protocols for Hospital Newborn Hearing Screening were completed and distributed in December 2002. Newborn Hearing Manuals were distributed to all birthing hospitals. Hospital trainings were conducted in December 2002 and March 2003.
 - The protocols for Audiological Diagnostic Assessment were completed and distributed in December 2001 to all of the licensed audiologists in TN. It was presented at the Tennessee Association of Audiology and Speech/Language Pathology 2002.
 - The protocols for early intervention follow-up and tracking were completed in May 2003. Training for Tennessee Early Intervention System (TEIS) (IDEA Part C) staff is scheduled for June 2003 and will be held in the three grand regions of the State.
 - The updated of list of Pediatric Hearing Screening, Audiologic Diagnostic and Early Intervention Providers was completed in April 2003. The document identifies pediatric providers, by region, that screen, diagnosis, fit hearing aids, provide cochlear implants and provide early intervention services. The document can be used by professionals and by families.
2. Develop and initiate a public awareness and training plan for professionals and consumers.
 - A training plan was developed in conjunction with the Newborn Hearing Screening HRSA grant.
 - Four brochures have been developed to increase public and professional awareness. The brochure titles are as follows: Your New Baby's Hearing (prenatal), Talking to Parents About Hearing Loss (professionals), Your Baby's Hearing Screening Suggests a Referral (infant that did not pass hospitals screen), and Why Bother With Newborn Hearing Screens and Hearing

Tests? (professionals and parents). The first two brochures will be available to the public in June 2003.

- Photographs of infants and families were completed in May 2003. They will be used for posters, brochures and for other media purposes.
- 95% of all newborns in Tennessee will receive a hearing screening prior to discharge or prior to one month of age.
- In April 2003, 82 of the 89 birthing facilities were conducting universal newborn hearing screenings. This indicated that 98% of the TN birth population had access to hearing screening. By July 2003, all but two small birthing hospitals with provide hearing screening.
- In April 2003, 57 of the 82 birthing centers that provided hearing screening also reported the results of the hearing screen to the State laboratory on the blood spot form. The 57 hospitals that reported indicated a 49.32% hearing-screening rate for 2002.
- 95% of children identified with a suspected hearing loss will receive an audiologic/diagnostic assessment prior to three months of age.
- In December 2002, only 25% of the infants received hearing assessment. The data was based on the number of infants reported on blood spot only.
- 95% of children diagnosed with a hearing loss will receive intervention services prior to six months of age.
- Tracking of intervention services will be captured by the TEIS data entry system. The file-maker system is to be implemented in July 2003.
- Parent and consumer participation will be evident in all aspects of program development.
- Ten parents of children identified with hearing loss participated in two panel discussions conducted at workshops for hospital and early intervention staff in regard to newborn hearing screening and tracking.
- Two parents attended the National EHDI workshop.
- The Newborn Hearing Screening Parent Consultant prepared newspaper and magazine articles on newborn hearing. The consultant prepared the four new brochures, in cooperation with NHS committee members.

METHODOLOGY:

- The Newborn Hearing Program is responsible for two grants related to the development of a sustainable early hearing, detection, and intervention surveillance system. A HRSA grant was awarded in March 2001 and a CDC grant was awarded in August 2001. These grants enabled the NHS program to develop reporting and data systems that are interactive with the Neometrics Genetics/Metabolic Screening, the Health Department PTBMIS System, the Health Department CIT System, and the Part C Early Intervention Data System.
- The data systems are linked and ready to process the new TEIS data collection for intervention and follow-up to be implemented in July 2003. The system will capture newborn hearing screening follow-up and track the diagnostic and intervention services provided for individual infants. The system will track types of providers and outcomes of language skills at one, two and three years of age.

COORDINATION:

- The NHS Task Force has here-to-fore implemented the same strategies for departmental and agency links that are planned through the Genetics Planning Grant. The NHS Program works

with local hospitals, Tennessee Hospital Association, Tennessee Perinatal Association, the Genetics Advisory Committee, Tennessee Chapter Family Voices, Tennessee Chapter AAP, Tennessee Association of Audiologist and Speech/Language Pathologists (TAASLP), Tennessee Early Intervention System (Part C IDEA), Tennessee Infant Parents Services (TIPS), Tennessee School for the Deaf, Tennessee Center on Deafness, Title V CSS Advisory Committee, Tennessee Services for the Deaf and Visually Impaired as well as others.

- In February 2003, Tennessee Newborn Hearing Screening representatives attended a National Annual Conference on Early Hearing Detection and Intervention sponsored by the CDC and HRSA. The TN team included CDC EHDI grant information systems staff, an audiologist, a parent of a child with hearing loss, the Family Voices –TN director, the TN AAP EHDI Chapter Champion, an early intervention Part C representative and the TN Newborn Screening director.
- The NHS program worked with representatives of the deaf and hard of hearing community to provide public education on newborn hearing screening.
- Funds are being provided to 22-24 small hospitals for the purchase of newborn hearing screening equipment. The Neometrics Newborn Screening System generated preliminary reports on the number of hearing screenings submitted on the blood spot form by the hospital. Results are reported to the hospital and medical provider on the blood spot laboratory mailer. Request for follow-up are conducted through the same system and in the same manner as the Genetic/Metabolic Newborn Screening. The NHS Task Force discussed the possibility of recommending proposed legislation for mandatory hearing screening be resubmitted to the legislature in 2003. Subcommittees were set up for Education/Training; EI Protocols; Hospital Screening Protocol, and the Genetics Planning Grant. Dr. Carmen Lozzio and Jacque Cundall will be the NHS Task Force representatives to the Genetics Planning Grant Committee.
- The results of activities are addressed in the Objectives section of this document. In addition, the pending revisions to TN Rules for Genetic and Metabolic Screening will require hospitals to report hearing screening and will clarify the role the Department of Education, IDEA Part C, Tennessee Early Intervention System (TEIS) will play in child find and early intervention tracking and follow-up.

EVALUATION AND MONITORING:

Evaluation and monitoring activities will be conducted as outlined in the Early Hearing Detection and Intervention CDC and HRSA Grants.

- The number hospitals providing and reporting newborn hearing screening is monitored through Neometrics system reports.
- The number and percent of infants referred for further screening will be monitored through the Neometrics system reports.
- The number of infants completing further hearing testing after referral and the results of the testing will be monitored by the Neometrics system and the TEIS data system.
- Infant profile data will be monitored through the TEIS data system and CIT.

Further information may be obtained by contacting the Tennessee Department of Health, Jacque Cundall, Newborn Hearing Screening Coordinator, HRSA grant program director (615-741-0310) or Dr. Richard Urbano, CDC EHDI grant program director (615-741-5001).

CONSUMER INVOLVEMENT SUBCOMMITTEE

MISSION STATEMENT:

- Ensure that families are educated, empowered and engaged in advocating for the health and related services needed for children who have or are at risk for chronic physical, developmental, behavioral or emotional conditions (Children with Special Health Care Needs, or CHSCN).
- Ensure that families are included as decision-making partners when accessing health and related services for their CHSCN.
- Ensure that families are satisfied with the quality of the health and related services for their CHSCN.

PROBLEM:

- Family difficulty in obtaining information about their child's medical condition, available resources and care services/choices.
- Overall lack of support from employers, school system, insurance programs and related services, including no "liaison person" for families of CHSCN.
- Overall disconnect between services and lack of coordination among the PCP, specialists, and therapists.
- Inadequate availability and access to mental health and home health services.

GOAL:

- Families will be able to access needed health and related services for their CHSCN, participate in decision making about their CHSCN when accessing services, and will be satisfied with the services they receive.

OBJECTIVES:

- Increase awareness of resources, providers and support groups for families and affected children, adolescents and adults.
- Inform consumers of anticipated additions to Tennessee newborn screening panel.
- Decrease the percent of families of CHSCN that report problems and/or are dissatisfied with services.

METHODOLOGY:

- Develop or coordinate development of a brochure with websites, support groups, etc., utilizing translators as necessary.

- Assist Tennessee Department of Health (TDH) in revising brochures, websites, educational presentations regarding expanded newborn screening panel, utilizing translators as necessary.
- Distribute information in Tennessee Genetic Outreach Clinics.
- Use results of Family Voices Federal Family-To-Family Grant focus groups to further define problems and develop strategies to improve satisfaction.
- Use results of periodic Family Voices Family-to-Family/Children's Special Services surveys to determine consumer satisfaction and identify issues for further intervention.
- Results of consumer focus groups and surveys will be presented to constituent advocacy and specialty services associations in newsletters, educational programs (PowerPoint) and other media.
- Post list of resources for families of CSHCN on TDH website.
- Track TennCare PCP utilization via TDH Children's Information Tennessee (CIT).
- Serve as a liaison with Tennessee Early Intervention System (TEIS), Children's Special Services (CSS), and school-based intervention service director to better coordinate services for families with CSHCN.

COORDINATION:

- Membership of the Consumer Involvement Subcommittee is comprised of stakeholders in the health and related services community, including family members of CSHCN and key advocacy organizations representing CSHCN.
- The Consumer Involvement Subcommittee will work collaboratively with the Family Voices Family-to-Family Program and Children's Special Services in designing satisfaction evaluation tools and communicating results of consumer surveys and focus groups.
- Member(s) of the Consumer Involvement Subcommittee will serve as liaison with TEIS, CSS and school-based intervention services.

EVALUATION AND MONITORING:

- Process will be evaluated by completion of products and tasks, tracking materials distributed, website hits, tracking the number of community presentations as well as the number of participants at community presentations.
- Process monitoring will be accomplished by review of objectives and activities at regular meetings of the Consumer Involvement Subcommittee.
- Outcome of advocacy empowerment efforts, participation in decision-making, and satisfaction with services will be evaluated by results of consumer focus groups and periodic consumer surveys.
- Participants at presentations will evaluate effectiveness of community presentations through analysis of assessment tools completed.

GENETICS PLAN IMPLEMENTATION:

PROPOSED ACTIVITIES:

Statewide Genetics Coordinating Committee and Subcommittees:

All the members of the Statewide Planning Committee have agreed to continue working as members of the SGCC. Representatives from other state departments have been invited to participate in this Committee and in the coordinating activities. A representative from the Bureau of TennCare (Tennessee Medicaid Program) has already been incorporated and a representative from the TN Department of Children's Services has been invited to participate. Interagency agreements have been signed by the representatives of the Departments already included in this Committee and will be signed by the new members. At the last meeting of the SGCC on May 9 a proposal was made to create a Legislative subcommittee to work with Tennessee Representatives and Senators concerning legislation related to NBS and NHS programs.

This Committee meets at least twice a year and reviews and approves the reports of the Subcommittees. Most of the activities to implement this plan will be accomplished by the subcommittees and are outlined in the attached reports. An Executive Committee (EC-SGCC) (co-directors, coordinator and/or program manager and chairs of subcommittees) oversees the work of the Subcommittees and the chairs report the EC-SGCC quarterly.

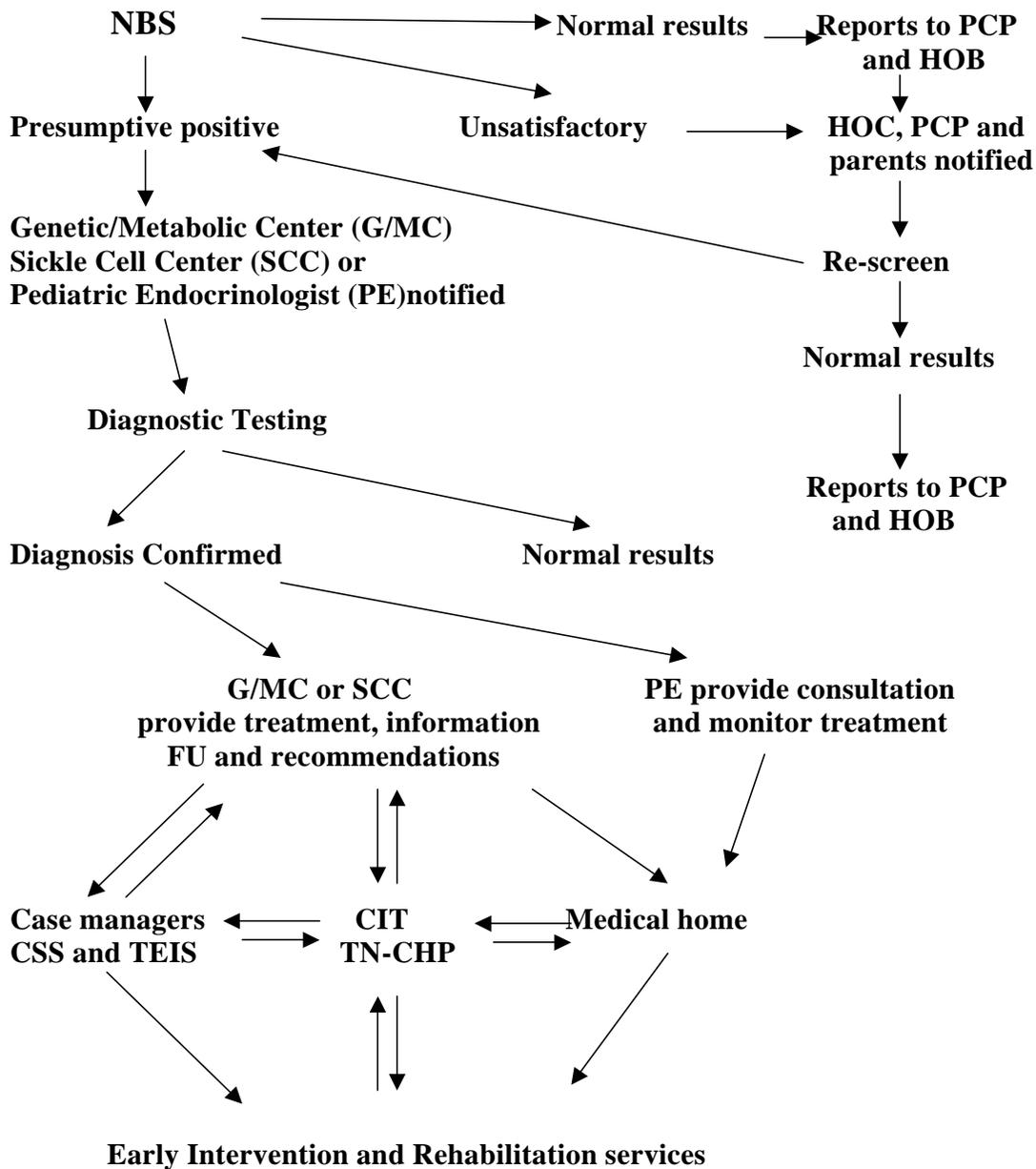
Integration of the State Genetics Plan and the State Information Technology Plan:

Tennessee is one of five states that has received funding in 2003 from HRSA Maternal and Child Health Bureau Joint Program Initiative between the Genetics Service Branch and the Office of Data Integration and Management for "Promoting Integration of State Information Systems and Newborn Screening Service Systems for Monitoring and Ensuring Quality Services to Newborns and Children With or At Risk for Heritable Disorders" This project is based on the implementation of the Genetics Plan and the IT Plan with strong cooperation and collaboration of all the members of the SGCC and subcommittees.

Regular and frequent conference calls and meetings of the three Co-Directors, Program Coordinator and/or Program Manager will be conducted to decide the specific activities required to achieve the goals of this Plan. Regular meetings and conference calls of the members of the subcommittees are also scheduled to implement the various aspects of the Genetics Plan. Special attention will be given to the integration of data incorporated into the TN-CHP with the NBS, NHS and other genetic services as well as with early intervention services.

The following Flow Charts show how Genetic services will be integrated to the Information Technology Plan:

SERVICE FLOW CHART



MONITORING AND EVALUATION

Details of the monitoring and evaluation process, tracking methods, outcomes, outputs, and products have been prepared for each goal and specific objectives. The epidemiological aspects of the program will be analyzed and utilized as an evaluative component. The TNCHP will provide a comprehensive, longitudinal picture of the characteristics of CSHCN and the services they receive and thus provide the foundation for outcome evaluation. Standard reports, analyses, and data visualization (maps, figures, charts) will delineate the array and magnitude of services available and accessible and the outcomes for the target population. Program staff will have the information needed to plan for policy changes directly affecting service delivery and outcomes. In addition to the use of systematically acquired data, results of Focus Groups and periodic surveys of consumers will be analyzed to develop strategies to improve outcomes.

The monitoring and evaluation for each specific goal and objectives will include the following:

1. Enhance and expand the state newborn screening program

The co-director for Genetic services and TDH staff will monitor the progress in the implementation of testing for additional disorders and the information on FU services for all disorders tested by the state laboratory. The progress made will be reported to the EC-SGCC monthly and to the SGCC and GAC at their bi-annual meetings of these committees.

The NBS, Ethics and Consumer Involvement subcommittees will evaluate the national recommendations of the HRSA-ACMG Expert Group and will report to the EC-SGCC and GAC. Information on the experience of other states will be reviewed and used to provide additional recommendations.

The health outcomes to be evaluated are:

1. Compare the number of presumptive positives and confirmed diagnoses for each disorder in the three regions of the state yearly.
2. Incorporate in the quarterly reports of Genetics and Sickle Cell Centers the number of children detected by NBS that are treated, receive genetic counseling and case management at these centers. Add this data to the TN-CHP and compare trends between regions and over time during the four years of the project.

2. Expand and enhance the public health information system infrastructure

The co-director for Health Information, the Epidemiologist/Program Manager and Data Manager will monitor and evaluate the progress made in adding data to the CIT system to create the TN-CHP and will report to the EC-SGCC. The subcommittees on Data, Ethics and Consumer Involvement will provide recommendations and the EC-SGCC will report to the SGCC and GAC. At the end of this project recommendations will be made to the Commissioner of TDH for continuation of the activities started by this program.

The outcomes to be monitored are:

1. Linkage of data on NBS and NHS to hospital of birth ID, heel stick card number, electronic birth records, deaths, WIC, immunizations, and other basic and enhanced CIT data (see IT Plan) will help determine incidence, morbidity and mortality of each disorder.
2. Reports, charts, graphs, maps and analyses will be made available through an enhanced web based query/display interface. For example, one page history of all screenings, diagnoses, treatments, medications and primary care providers (PCPs) will be available for each child to authorized providers. The number of times this page is accessed and by whom will be recorded for further statistical analyses.

3. Integrate systems of early identification with early intervention

The EC-SGCC will evaluate the recommendations from the Ethics and Consumer Involvement subcommittees concerning issues of confidentiality, informed consent, access to medical records and other ethical, legal and privacy issues. Additional topics that will be considered include recommendations to further the agenda for Healthy People 2010, improving service delivery to women, children youth and families from communities with limited access to comprehensive care. These recommendations will be presented to the SGCC and feedback from this committee will be used to develop additional recommendation. The members of the SGCC representing different program of TDH-MCH will make recommendations on how the SGCC will develop measures that will complement current Title V Block Grant Performance measurements and the SGCC will forward these recommendations to the director of MCH. Monitoring and evaluation of these activities will continue during the duration of the project.

Monitoring and evaluation of coordination of services at the local level will be based on results of surveys and Focus groups with consumers. The Genetic Counselor will work with the Consumer Involvement subcommittee and Family Voices of TN to conduct these efforts. The results of these surveys and Focus groups will be reported to the EC-SGCC and SGCC and plans to improve coordination of services will be based on measures to address the gaps and deficiencies reported.

Monitoring and evaluation of linkage of data to coordination of services is described in the following section.

The outcomes to be evaluated are:

1. Data on referrals of infants diagnosed through NBS or NHS to case management and/or early intervention services such as CSS and TEIS will be added to the quarterly reports of the Genetics and Sickle Cell Centers and included in the CHP. This data will be analyzed yearly to assess increases in the rates of referrals as the results of improvement in coordination of services.
2. Surveys and Focus groups with consumers will be conducted in collaboration with Family Voices of TN, interviews with case managers and data from TN-CHP will be used to assess progress towards a comprehensive continuum of services for infants and children with disorders detected by the NBS program.

4. Implement quality assurance and evaluation of service delivery systems to determine system effectiveness and efficiency

The Data Manager, Epidemiologist/Program Manger and the Genetic Counselor will monitor and evaluate these activities. The progress made will be reported to the Data subcommittee, the EC-SGCC and the SGCC.

The following outcomes will be monitored:

1. The integrated data in the TN-CHP will be used to track the progress through the system, identify gaps in services and generate reports.
2. Reports will be analyzed and used for evaluation of services and for planning of future activities.

5. Increase provider's understanding of the importance of timely and appropriate treatment for children with genetic conditions

The Professional Education subcommittee has developed a specific plan described in the attached report and Work Plan Matrix. The Genetic Counselor will work with the members of this subcommittee to accomplish the proposed activities and monitor the progress made. The Chair will report to the EC-SGAC and the SGCC of the achievements towards this goal. Special efforts will be made to make available to professionals and consumers culturally sensitive materials translated to the most common languages of residents of TN.

The achievement of this goal will be monitored as follows:

Increase health care professional's awareness of all aspects of disorders detected by NBS. Surveys will be prepared and sent yearly to assess level of knowledge before and after distribution of printed material, web-based information, videos, and other educational programs.

6. Empower families to be effective advocates for appropriate care and services for their CSHCN

The Consumer Involvement subcommittee has developed a specific Plan of activities aimed at the goals proposed by the agenda of Healthy People 2010. The details of this Plan are reported in the Report of this subcommittee. The members of this subcommittee will discuss all aspects of this plan at bimonthly or monthly conference calls and will report to the EC-SGCC and SGCC.

The outcomes to be evaluated are:

1. Periodic surveys of parents to assess their understanding of the care their child is receiving and to learn about any unmet concerns about care and/or data management.
2. Assess the frequency with which parents access their child CHP and the state website.
3. Assess the data security and confidentiality of the CHP.
4. Effectiveness of community presentations will be evaluated through analysis of assessment tools completed.

The above stated evaluations and monitoring of outcomes are preliminary and will be reviewed and revised as results of the initial assessments reveal areas of strength and weakness.