Newborn screening is recognized internationally as an essential, preventive public health program for early identification of disorders in newborns that can affect their long term health. Early detection, diagnosis, and treatment of certain genetic, metabolic, or infectious congenital disorders can lead to significant reductions of death, disease, and associated disabilities” (National Newborn Screening and Global Resource Center, 2012)

Program Purpose

The Connecticut Newborn Screening (CT NBS) Program works to assure that every newborn, who is born or resides in Connecticut, has a valid newborn screening on record and that those infants with abnormal screening results are promptly referred to a specialty treatment center for further evaluation and treatment when needed. These comprehensive efforts help prevent unnecessary disability and premature death.

History & Description

In 1964, the CT Newborn Screening (NBS) Program began statewide blood-spot screening for two disorders, Phenylketonuria (PKU) and Galactosemia (GALT). Between 1976 and 1993, many disorders were added to the screening CT NBS panel, including Congenital Hypothyroidism (CH), Congenital Adrenal (CAH), Maple Syrup Urine Disease (MSUD), Homocystinuria (HCY) and Biotinidase Deficiency (BIO), and Hemoglobinopathies, such as Sickle Cell disease (SCD), to name a few. In May 2004, CT NBS program implemented Tandem Mass Spectrometry (TMS) instrumentation. This technology allowed the lab to screen for over 60 conditions and disorders that include Amino Acid (AA), Urea Cycle (UCD), Fatty Acid Oxidation (FAO), and Organic Acid (OA) disorders, all from 5 small blood-spots. Screening for Severe Combined Immune Deficiency (SCID) was added in 2011 and most recently, in early 2016, the lab began screening for Adrenoleukodystrophy (X-ALD).
The Connecticut Department of Public Health (CT DPH) Regulations (Sec.19a-55-1-19a-55-3) require those overseeing the medical care of newborns to collect a blood-spot specimen, from each newborn infant in their care, for genetic and metabolic testing as prescribed by the CT DPH, in accordance with Sec. 19a-55 of the Connecticut General Statutes. This specimen must be collected before the fourth day of life (96 hours of age), prior to discharge from the birthing center/birth hospital or as soon as medically appropriate after birth. This blood-spot specimen must be shipped to the State Laboratory or designated facility within 48 hours of collection. Please refer to the CT NBS Specimen Collection Guidelines for specific recommendations on the timing of and procedure for NBS specimen collection. Birthing facilities, NICU care providers, midwives and primary care providers have a responsibility to know the NBS status of each infant in their care, and to assure that each infant has a valid newborn screen on record. This includes infants who may not have been born in the state but reside and/or receive medical care in the state.

Connecticut State Statute also mandates newborn screening for Cystic Fibrosis (CF), hearing deficits, serious heart defects (Critical Coronary Heart Defects—CCHD) and HIV (Human Immunodeficiency Virus). Cystic Fibrosis testing is conducted by the CF programs of Yale-New Haven Hospital (YNHH) and the UCONN Health Center (UCHC). Screenings for hearing, CCHD and HIV (Human Immunodeficiency Virus) are usually conducted by the hospital of birth within the first few days of life.

**Testing, Tracking (Short-Term Follow-Up) and Treatment**

The CT NBS program employs Nurse Consultants who work with CT birth hospitals, birthing centers and midwife groups to monitor births through the NBS database (also known as Maven or CT-Site) and assure that the State Laboratory has received a satisfactory NBS blood-spot specimen for every newborn. The nurse consultants also work to identify babies residing in CT, who are not in the NBS database (i.e.: babies born out of state, adopted and babies born outside of the hospital—such as home births and other out of hospital births that are not attended by a licensed medical/midwife provider). These nurses also follow-up on specimens received that are unsatisfactory or invalid for testing. All NBS specimens undergo extensive testing for over 60 conditions at the Connecticut State Laboratory. The Connecticut newborn screening panel is based on recommendations set forth by the US Department of Health and Human Services (US DHHS) and includes testing for metabolic disorders such as amino acid fatty acid oxidation, and organic acid disorders, in addition to, endocrine, immunodeficiency, and hemoglobin disorders.

The nurse consultants follow-up on all abnormal NBS results and either request a specimen for repeat NBS analysis or refer the child to a specialist in genetics, endocrinology, hematology or immunology at one of three regional treatment centers. The treatment center specialists work with the hospital of birth, PCP, Midwife or NICU to provide comprehensive care that includes consultation, diagnostic testing, education, counselling and treatment when indicated. Short-
term follow-up by the NBS program continues until the child receives a confirmed diagnosis or a condition is ruled-out.

In addition to the work previously described, CT NBS Program staff also collect and analyze data that are used for quality assurance activities, epidemiological reports and to advance the science of NBS.