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University of Connecticut Health Center

Department of Genetics and Developmental Biology
Division of Human Genetics
and the
School of Medicine
Office of Continuing Education
and the

CONNECTICUT DEPARTMENT OF PUBLIC HEALTH

Keeping Connecticut Healthy

Family Health Section
Public Health Initiatives Branch
J. Robert Galvin, MD, MPH
Commissioner

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Newborn Screening in Connecticut

WEB-BASED TRAINING

for Pediatricians, Family Practitioners, Advanced Practice Nurses and Physician Assistants

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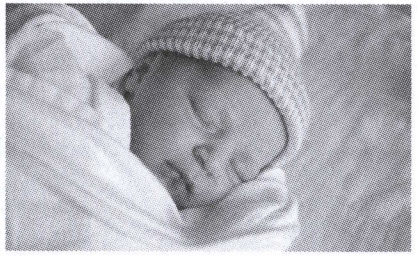

Each year, the Connecticut Department of Public Health's Newborn Screening Program identifies more than 2,100 infants with a potential genetic disorder or permanent congenital hearing loss.

To register, go to:
www.genetrain.org

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- ***Newborn Screening in Connecticut*** **features the state's foremost experts on newborn screening disorders.**
- **The training is comprised of 7 individual sessions, each approximately 30 minutes in length.**
- **Participants can complete each session separately and return to the training at their own pace.**

Learning Objectives

Session 1:
Introduction to Connecticut's Newborn Screening Program

Robert M. Greenstein, MD
Division of Human Genetics
UCONN Health Center

Learning Objectives: 1) Explain the history and evolution of the Connecticut Newborn Screening Program (NBSP). 2) Describe how this program is guided by the "3 T's" - Testing, Tracking and Treatment. 3) Describe the major categories of disorders in the NBSP. 4) Explain how the medical home interacts with the NBSP and the family.

Session 2:
Amino Acid, Organic Acid & Fat Oxidative Disorders

Margretta R. Seashore, MD
Yale Department of Human Genetics
Yale University School of Medicine

Learning Objectives: 1) Explain the basic principles of diagnosis and treatment of inborn errors of metabolism. 2) List the amino acid, organic acid and fatty acid metabolism disorders identified in Connecticut's NBSP. 3) Describe the strategies for responding to a positive newborn screening test. 4) Identify the role of the primary care provider in newborn screening.

Session 3:
Congenital Hypothyroidism and Congenital Adrenal Hyperplasia

Scott A. Rivkees, MD
Department of Pediatrics
Yale University School of Medicine

Learning Objectives: 1) Describe the evaluation and treatment of infants with a positive newborn screening test for congenital hypothyroidism. 2) Describe the evaluation and treatment of infants with a positive newborn screening test for congenital adrenal hyperplasia. 3) Explain the role of the primary care provider when there is a positive test result for congenital hypothyroidism or congenital adrenal hyperplasia.

Session 4:
Newborn Screening for Hemoglobinopathies

Howard A. Pearson, MD
Director, Yale Sickle Cell Service
Yale University School of Medicine

Learning Objectives: 1) List the major reasons for neonatal testing for hemoglobinopathies. 2) Explain the preferred methods for neonatal testing for hemoglobinopathies. 3) Describe the neonatal hemoglobin patterns of normal infants, sickle cell trait, sickle cell anemia, and hemoglobin SC disease. 4) Describe the screening and management protocols for hemoglobinopathies in Connecticut.

Session 5:
Newborn Screening for Cystic Fibrosis

Sidney M. Hopfer, PhD
Department of Laboratory Medicine
UCONN Health Center

Learning Objectives: 1) Explain the protocol used to screen for cystic fibrosis (CF) in newborns. 2) Review the follow-through process between the screening lab, CF physician, pediatrician and infant's family.

Session 6:
Early Hearing Detection & Intervention

Donna C. Maselli, RN, MPH
Connecticut Department of Public Health

Learning Objectives: 1) Discuss two benefits of early intervention for children with hearing loss. 2) Recall that not all hearing loss occurs at birth. 3) Describe risk indicators for hearing loss that require more frequent monitoring. 4) Explain the role of the medical home in the early hearing detection and intervention process.

Session 7:
Genetic Testing for Infants with Hearing Loss

Sally S. Rosengren, MD
Division of Human Genetics
UCONN Health Center

Learning Objectives: 1) Recognize that most cases of newborn and childhood hearing loss are genetic. 2) Differentiate between syndromic and non-syndromic hearing loss. 3) List the benefits and limitations of genetic testing for hearing loss. 4) Recognize the most common hearing loss genes and syndromes.