Newborn Screening Follow Up Tandem Mass Spectrometry Workshop
January 13-17, 2020
Silver Spring, MD

Course Description:
This intensive five-day course assumes a basic understanding of newborn screening follow-up and metabolic biochemistry. Coursework will begin with a review of the principles of tandem mass spectrometry, diagnostic patterns in results, cut-offs, biochemical pathways, diagnostic follow-up and biochemical and clinical features of the metabolic disorders. Each day will cover interpretive skills and diagnostic follow up of certain disorders detectable through MS/MS screening including: amino acid disorders, urea cycle disorders, fatty acid oxidation disorders, and organic acid disorders. Interpretation homework assignments will be given along with daily examinations of information learned.

Level of Instruction:
Intermediate

Audience:
This program is intended for US-based newborn screening follow-up staff.

Faculty:
Dr. David Millington, Professor Emeritus of Pediatrics at Duke University School of Medicine.

Prerequisites
• Applicants should be actively engaged in follow-up activities in US-based newborn screening programs.
• Applicants must have a basic understanding of newborn screening follow-up and metabolic chemistry.

Application:
• Apply Here by September 29, 2019

Learning Objectives:
• Interpret the results obtained from MS/MS analysis of dried blood spots for:
  o Amino Acidopathies and Urea Cycle Disorders.
  o Fatty acid oxidation disorders
  o Organic acid disorders.
• Identify appropriate expected ranges and cutoffs for MS/MS when applied to newborn screening disorders
• Describe the biochemical and clinical features of the metabolic disorders
• Recommend appropriate follow-up tests for confirmation of screening results and differential diagnosis

Learning Goals:
• Describe MS/MS as related to clinical diagnostics and NBS.
• Describe the basic theory of ESI and MS/MS.
• Describe scan functions and how they are employed in NBS.
Describe the biochemistry and clinical manifestations of amino acid and urea cycle disorders and the source of abnormal metabolites in disease states.

Describe the biochemistry and clinical manifestations of disorders of the catabolism of branched-chain amino acids and related disorders that comprise the organic acidurias.

Describe the biochemistry and clinical manifestations of inherited disorders of mitochondrial fatty acid beta-oxidation and the origin of diagnostic metabolites.

Describe roles of personnel required for NBS expanded with MS/MS.

Interpret amino acid MS/MS spectra and diagnose amino acid disorders.

Identify disorders of amino acid catabolism by their acylcarnitine spectra and understand confirmatory test procedure.

Identify disorders of fatty acid oxidation by their acylcarnitine spectra and understand confirmatory test procedure.

Describe the meaning of stable isotopes and their role in quantitative MS.

Describe how cut-offs are established and affect result reporting.

Summarize implementation of expanded NBS in the state of New York.

Summarize the impact of expanded newborn screening from the genetic counselor’s point of view.

Summarize a dietitian’s role in follow-up of patients identified by NBS.

Describe response to abnormal NBS results: follow-up testing of amino acids, organic acids and acylcarnitines for diagnosis – limitations.

Describe how disorders are selected for the panel.

Describe second-tier follow-up testing available by MS/MS methods.

Describe the implementation of MS/MS in a State NBS Program – review of problems identified and their resolution, interesting case reports.

Understand the impact of newborn screening in North Carolina – disorders detected and frequency.

Identify non-diagnostic MS/MS results and understand causes.

Summarize the CDC QA/QC program for expanded newborn screening.

Describe how in vitro tests are used to confirm fatty acid oxidation defects.

Describe how new technologies will enable further expansion of newborn screening for lysosomal storage disorders (LSD) and severe combined immune deficiency (SCID).

Interpret and respond to abnormal MS/MS screening results.

The Association of Public Health Laboratories (APHL) is approved as a provider of continuing education programs in the clinical laboratory sciences by the ASCLS P.A.C.E.® Program. Participants who successfully complete this program will be awarded 29.5 contact hours.

Staff Contact:
Erin Darby
erin.darby@aphl.org