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# 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:
  - Amino Acidopathies and Urea Cycle Disorders.
  - Fatty acid oxidation disorders
  - 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.

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- 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