2026 Newborn Screening and Genetics

Tandem Mass Spectrometry Follow-Up Workshop

August 10 – 14, 2026

APHL Headquarters
7700 Wisconsin Avenue, Suite 1000
Bethesda, MD 20814

This meeting is sponsored by the Association of Public Health Laboratories

Purpose

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 (MS/MS), diagnostic patterns in results, cut-offs, biochemical pathways, diagnostic follow-up and biochemical and clinical features of 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, organic acid disorders, peroxisomal and lysosomal disorders.

The course content will be shared through didactic lectures and group discussions. Students learned knowledge will be assessed through quizzes and homework assignments. Successful course performance will be recognized with a certificate of course completion and continuing education units will be available.

Objectives

After completing this workshop, participants will be able to 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, peroxisomal and lysosomal disorders; perform a risk assessment based upon the analyte values and expected ranges, along with demographic information; describe the biochemical and clinical features of the metabolic disorders; learn the appropriate follow-up tests for confirmation of screening results and differential diagnosis; and understand the importance of continued long-term follow-up after diagnosis.

Disclosures

There is neither presence of conflict of interest nor commercial and sponsorship support among planners, faculty, or moderators for this workshop.

Non-Endorsement of Products

As an Accredited Provider of Continuing Education (CE) Credits for Health Education (CHES), Nursing (CNE), Medicine (CME), and Public Health (CPH) the American Public Health Association Center for Professional Development (CPD) Unit and its respective Continuing Education accrediting organizations do not endorse any products or services that are displayed or referred to in conjunction with this activity and are not responsible for the actual presentation of content during scientific sessions.

Policy on Disclosures to Learners

The Association of Public Health Laboratories strives to ensure balance, independence, objectivity and scientific rigor in all of its educational programs. All planners, faculty members, moderators, discussants, panelist and presenters participating in this program have been required to disclose any real or apparent conflict(s) of interest that may have a direct bearing on the subject matter of this program. This includes relationships with pharmaceutical companies, biomedical device manufacturers or other corporations whose products or services are related to the subject matter of the presentation topic. The intent of this policy is to identify openly any conflict of interest so that the attendees may form their own judgments about the presentation with full disclosure of the facts. In addition, faculty is expected to openly disclose any off-label, experimental and/or investigational uses of drugs or devices in their presentation. Disclosures, Conflict of Interest (COI) and Resolution of COI policies are available via the APHA's website and in the printed program.

Nursing (CNE) Accreditation

This nursing continuing professional development activity was approved by the American Public Health Association's Public Health Nursing Section Approver Unit, an accredited approver by the American Nurses Credentialing Center's Commission on Accreditation.

Consent to Use Photographic Images

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Well-Being

To ensure the health and well-being of everyone participating in this meeting, we kindly remind you to prioritize your health and that of others when deciding whether to attend in person meetings, especially if you are feeling unwell.

If you are experiencing any symptoms of illness or have been in contact with someone who is sick, we strongly encourage you to participate remotely or to reschedule your participation in the meeting. Your health and safety are our top priorities, and we appreciate your cooperation in maintaining a healthy work environment for all.

Contact

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AGENDA

Day 1	Monday | August 10, 2026
8:30 – 9:00 am 30 minutes	**Arrival and Breakfast**
9:00 – 9:30 am 30 minutes	**Welcome, Opening Remarks, and Introductions** Scott Becker, MS, Chief Executive Officer, APHL Jelili Ojodu, MPH, Senior Director, Newborn Screening & Genetics, APHL Patrice K. Held, PhD, PhD FACMG, Newborn Screening Program Manager, Oregon State Public Health Laboratory
9:30 – 9:45 am 15 minutes	**Pre-Assessment and Review** Review
9:45 – 10:15 am 30 minutes	**Brief History of Newborn Screening (NBS) and How It Was Transformed By Tandem Mass Spectrometry (MS/MS)** Lecture
10:15 – 10:45 am 30 minutes	**Fundamentals of Liquid Chromatography (LC) and MS/MS** Lecture • How does the MS/MS work? • Different scan functions to look for amino acids (AA), acylcarnitines (AC), and other analytes.
10:45 – 11:00 am 15 minutes	**Break**
11:00 – 11:30 am 30 minutes	**Analyte Quantitation in NBS Using MS/MS** Lecture • How analyte values are calculated. • Use of stable isotopes
11:30 – 12:00 am 30 minutes	**From Punch to Analysis: Laboratory Workflow** Lecture • Step by step description of the common laboratory workflows for amino acid (AA) & acylcarnitine (AC) analysis
12:00 – 1:00 pm 60 minutes	**Lunch**
1:00 – 2:00 pm 60 minutes	**Basis of Metabolism** Lecture • In fed and fasting states, how and when does the body metabolize protein, carbohydrates, and fats?

2:00 – 2:30 pm 30 minutes	**Metabolism and Diet** Lecture • Discussion of metabolism and diet; ways in which metabolism is altered by dietary changes
2:30 – 3:00 pm 30 minutes	**Review of Central Dogma and Inheritance Patterns for Genetic Conditions** Lecture • Central dogma, correlating the deoxyribonucleic acid (DNA) to the protein definition to the elevation of analytes measured in the laboratory • Autosomal recessive (AR) and X-Linked inheritance patterns
3:00 – 3:15 pm 15 minutes	**Break**
3:15 – 4:00 pm 45 minutes	**Tell Me About Your Program: Testing Algorithms and Reporting Workflows** Discussion
4:00 – 4:30 pm 30 minutes	**Lecture and Discussion Recap** Review • Review concepts presented • Question and answer
6:00 pm	**Group Dinner** Sponsored by Association of Public Health Laboratories
Day 2	**Tuesday | August 11, 2026**
8:30 – 9:00 am 60 minutes	**Breakfast**
9:00 – 10:30 am 90 minutes	**Amino Acid (AA) and Urea Cycle Disorders** Lecture • All Recommended Uniform Screening Panel (RUSP) conditions including guanidinoacetate methyltransferase (GAMT) deficiency and ornithine transcarbamylase (OTC) deficiency • Pathway, clinical presentation, treatment • NBS profile review of conditions presented (1st and 2nd tier)
10:30 – 10:45 am 15 minutes	**Break**
10:45 – 12:00 pm 75 minutes	**Organic Acidemia (OA) Disorders** Lecture • All RUSP conditions • Pathway, clinical presentation, treatment • Newborn screening profile review of conditions presented (1st and 2nd tier)

12:00 – 1:00 pm 60 minutes	**Lunch**
1:00 – 2:00 pm 60 minutes	**Fatty Acid Oxidation (FAO) Disorders and the Carnitine Cycle** — Lecture • All RUSP conditions • Pathway, clinical presentation, treatment • Newborn screening profile review of conditions presented (1st and 2nd tier)
2:00 – 2:30 pm 30 minutes	**Lecture and Discussion Recap** — Review • Review concepts presented • Question and Answer
2:30 – 2:45 pm 15 minutes	**Break**
2:45 – 3:30 pm 45 minutes	**Timeliness in NBS and Importance of risk assessment** — Lecture • Why is timeliness important? • Define risk assessment (borderline versus panic)
3:30 – 4:00 pm 30 minutes	**Confirmatory Testing and Basic Laboratory Tests** — Lecture • Diagnostic testing performed in the clinical biochemical genetics laboratory • Plasma AA, urine OA, plasma AC, and other analyte specific testing
4:00 – 4:30 pm 30 minutes	**Case Presentations and Clinical Vignettes** — Discussion
Day 3	**Wednesday | August 12, 2026**
8:30 – 9:00 am 30 minutes	**Breakfast**
9:00 – 9:45 am 45 minutes	**Lysosomal Disorders** — Lecture • Role of the lysosome • Metabolism of macromolecules • Pompe, MPS I, MPS II, Krabbe, MLD • Pathway, clinical presentation (mild, severe, late onset) treatment
9:45 – 10:30 am 45 minutes	**Screening for Lysosomal Disorders** — Lecture • Step-by-step description of the common laboratory workflow for lysosomal disorders • Enzyme activity assessment combined with higher-tier testing, either molecular or biochemical testing • How to handle pseudo deficiencies and variants of unknown significance (VUS)

10:30 – 10:45 am 15 minutes	**Break**
10:45 – 11:30 am 45 minutes	**Screening for X-linked adrenoleukodystrophy (XALD)** Lecture • Role of the peroxisome • Pathway, clinical, presentation, treatment
11:30 – 12:00 pm 60 minutes	**Premature Babies and Influence on Profiles** Lecture • Influence of antibiotics, dietary interventions, other neonatal conditions
12:00 – 12:30 pm 30 minutes	**Result Review and Profiles** Discussion • Practice reviewing profiles, assessing risk
12:30 pm	**Free Afternoon**
Day 4	**Thursday | August 13, 2026**
8:30 – 9:00 am 60 minutes	**Breakfast**
9:00 – 10:30 am 90 minutes	**Long-Term Follow-Up (LTFU)** Lecture • Share experiences of states who are actively engaged in LTFU • Definitions of and barriers to LTFU • Address where short-term follow-up (STFU) stops and LTFU starts, especially for late onset conditions. • NewSTEPs LTFU Workgroup, Review of process measures and the Feasibility Pilot Study
10:30 – 11:00 am 30 minutes	**Tell Me About Your Program: Long-Term Follow-Up (LTFU)** Discussion • What successes has your state LTFU program achieved? • What challenges has your program encountered?
11:00 – 11:15 am 15 minutes	**Break**
11:15 – 12:15 pm 60 minutes	**Downstream Clinical Implications of Newborn Screening: A Genetic Counselor's Perspective** Lecture • Example cases: X-linked conditions, false negatives, maternal conditions • Social determinants of health and how it impacts follow-up care
12:15 – 1:00 pm 45 minutes	**Lunch**

1:00 – 2:00 pm 60 minutes	**Uncertainty in NBS and the Experience of Families**	Lecture
2:00 – 2:45 pm 45 minutes	**Tell Me About Your Program: Result Notification and Communication** • Share your experiences of uncertainty in NBS • Discuss challenges and barriers in reporting and communicating results	Discussion
2:45 – 3:00 pm 15 minutes	**Break**	
3:00 – 3:30 pm 30 minutes	**On the Horizon** • New conditions, increase in multiplexing and higher throughputs • Machine learning/artificial intelligence (AI) for data analysis • Research Initiatives (i.e., Screen Plus)	Lecture
3:30 – 4:00 pm 30 minutes	**Role of the Centers for Disease Control and Prevention (CDC) in Support of NBS programs**	Lecture
4:00 – 4:30 pm 30 minutes	**Ask the Expert**	Discussion
Day 5	**Friday | August 14, 2026**	
8:30 – 9:00 am 30 minutes	**Breakfast**	
9:00 – 10:00 am 60 minutes	**Quality Assurance (QA) within NBS Programs** • Laboratory quality assurance (QA) activities • Clinical Laboratory Improvement Amendments (CLIA) Standards and College of American Pathologists (CAP) Accreditation • Guidance from the Clinical Laboratory Standards Institute (CLSI) • Defining cutoffs and cutoff review • Investigation of false negatives	Lecture
10:00 – 11:00 am 60 minutes	**NewSTEPs Quality Assurance (QA) within NBS Follow-Up** • NewSTEPs Specimen Metrics • NewSTEPs Case Metrics • NewSTEPs Screen Positive Study	Lecture
11:00 – 11:15 am 15 minutes	**Break**	

11:15 – 11:45 pm 30 minutes	**Tell Me About Your Program: Quality Assurance and Management** Discussion • What quality assurance practices are utilized in your laboratory? • Share your quality management plan
11:45 – 1:00 pm 75 minutes	**Post-Test and Recap** Review • Boxed Lunch • Review of key concepts
1:00 pm	**Adjourn**