

NACB Laboratory Medicine Practice Guidelines
Expanded Newborn Screening for Metabolic Diseases by Tandem Mass Spectrometry and Follow-Up Testing
Chair: Michael J. Bennett PhD, FACB

Sub-Committees

1. Evidence-Based Rationale for Expanded Newborn Screening.

Goals: To bring together all evidence-based data to support expansion of newborn screening for metabolic diseases: Includes previously published reports, surveys and recommendations from ACMG, CDC, CLSI and other mandated and collegial organizations. Including International Inputs?

Chair: Piero Rinaldo MD, PhD, FACMG.

Members: Ronald J. Whitley PhD, FACB, Bill Rhead MD, PhD, FACMG, W. Harry Hannon PhD

2. Preanalytical, Analytical and Post-Analytical Issues Related to Tandem Mass Spectrometry as a Tool for Expanded Newborn Screening.

Goals: To define standards of acceptability for the whole process. Includes all preanalytical variables including process interference, the analytical process including correct use of internal standards, QC, performance evaluation (PT, CQI) and interpretation of data.

Chair: W. Harry Hannon

Members: Dennis Dietzen PhD, DABCC, Ronald J. Whitley, Piero Rinaldo

3. Follow-up of Positive Screens

Goals: To define acceptable limits for sensitivity and specificity for Tandem Mass Spectrometry. To define the appropriate follow up investigation of all cases that are positive

Chair: Uttam Garg, PhD, FACB

Members: Stanley F. Lo PhD, FACB, Bill Rhead, Dennis Dietzen

4. Patient Outcomes from Expanded Newborn Screening

Goals: To define patient benefits from early diagnosis as outcome data is starting to appear in the literature and pool outcomes data from multiple centers.

Chair: Bill Rhead

Members: Stanley F. Lo, Joe Muenzer MD, Piero Rinaldo

5. Future Directions in Expanded Newborn Screening for Metabolic Diseases

Goals: To bring together recent developments in Tandem Mass spectrometry for additional metabolic diseases including lysosomal storage and creatine disorders. This session will address evidence-based studies for future direction. May not become part of the final LMPG but is important for total picture.

Chair: Joe Muenzer

Members: Dennis Dietzen, Bill Rhead, Uttam Garg