

LABORATORY STANDARDS AND PROCEDURES

WORKGROUP: REVIEW OF THE OVERVIEW OF RISK DETERMINATION DOCUMENT

Presented to the Advisory Committee on Heritable
Disorders in Newborns and Children

February 8, 2018

Co-chairs: Kellie Kelm, PhD & Susan Tanksley, PhD

Timeline: Committee discussions on risk determination

1. Presentations at ACHDNC meetings in 2017

- February – Michele Caggana (NY), John Thompson (WA), Carol Johnson (IA)
- May – Michele Caggana (NY), Scott Shone (NJ), Amy Gaviglio (MN), also R4S and CLIR tools by Piero Rinaldo (Mayo Clinic) and CDC's NBS QA/QC Program by Carla Cuthbert (CDC)
- August – Susan Tanksley (APHL)

2. August and November - Discussion in the Workgroup



Analysis. Answers. Action.

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NBS QA/QC Subcommittee: Guidelines for Determining “Cutoffs”

Presentation to ACHDNC Laboratory Standards and Procedures
Workgroup

August 3, 2017

Presenters:

Joseph Orsini, Ph.D.

Patricia Hunt

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APHL presentations on risk determination document to the Workgroup

1. August 2017 – document outline presented
2. November 2017 – draft document provided for review
 - Workgroup provided feedback on both dates
3. January 2018 - document distributed to the newborn screening community in January for review and input
 - Most of the Workgroup's suggestions were addressed
4. January 2018 - Workgroup offered additional feedback regarding points of clarification

Workgroup discussion and conclusions:

1. The APHL document:

- describes the scientific processes that states currently use to determine which specimens test within normal range vs. out of range.
- is a valuable resources for state newborn screening programs.

1. The APHL document does not:

- include best practices for screening for all conditions.
- harmonize newborn screening tests across states.

2. APHL intends for this to be a living document that is revised over time.