



# Ad-Hoc Workgroup: Interpreting Newborn Screening Results Updates

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# Ad-Hoc Workgroup Members

- ▶ Mei Baker, Chair
- ▶ Cindy Powell
- ▶ Scott Shone
- ▶ Beth Tarini
- ▶ Sue Berry
- ▶ Susan Tanksley
- ▶ Natasha Bonhomme
- ▶ Debra Freedenberg
- ▶ Amy Gaviglio
- ▶ Joyce Graff
- ▶ Jeremy Penn
- ▶ Kyle Brothers

# Approach

- ▶ Report to the Committee
- ▶ Publication in Peer Reviewed Journal
- ▶ Slide deck or other tools for clinicians

# Report Structure

- ▶ Part one: Introduction
- ▶ Part two: Current Practice
- ▶ Part three: Discussion and Suggestions/Recommendations

# Part One—Introduction

## ▶ Rationale and targeted audience (family focus)

- ▶ The transparency of NBS's benefit and limitation is needed for health providers/organizations, parents, and public to set a realistic expectation.
- ▶ Consistent terminology is needed for health providers/organizations, parents, and public to understand what NBS is and is not

## ▶ Knowledge gap and unattainable expectations

- ▶ General medical screening concept
- ▶ Additional unique NBS aspects (newborns, time critical conditions, technologies, etc.)
- ▶ Difference between screening and diagnosis
- ▶ Terminology clarification

# Part One—Introduction

- ▶ **Screening test versus diagnostic test**  
references in literature; MOC from the Midwest Genetics Network; CLSI documents; CDC/APHL QA committee documents
- ▶ **Individual versus population (individual circumstance)**

# Part Two—Current Practice

## ▶ Risk assessment evidence

- ▶ Results are threshold based and categorical
- ▶ Results interpretation (risk for..., indication of...)
- ▶ Recommendation (further confirmatory testing and clinical assessment)

# Part Two—Current Practice

- ▶ **Report categories with emphasis on the associated actions**
  - ▶ Further action needed, with recommendation of confirmatory testing (screening positive; abnormal screening; out-of-range results)
  - ▶ Further action needed, with recommendation to repeat NBS (possible screening positive; possible abnormal screening; borderline results)
  - ▶ No further action needed unless clinically indicated (screening negative; normal screening; in-range results)
  - ▶ Unsatisfactory specimens with recommendation to collect new NBS specimen (inconclusive results, undermined results)
  - ▶ Results pending in two screening protocol



# Part Three—Discussion and Recommendations/Suggestions

- ▶ **Make NBS risk assessment more clear**
  - More explicit “risk assessment” language and methods for interpreting NBS results
  - Add on NBS interpretation for “normal NBS results”
- ▶ **Terminology clarification and consistency**
- ▶ **Strategies for communicating NBS results to families**
- ▶ **Strategies for improving NBS performance**

# Timelines

- ▶ ACHDNC meeting on March 22, 2019
  - ▶ Draft work plan - Committee feedback
- ▶ ACHDNC meeting on Apr 23-24, 2019
  - ▶ Outline of the report and *dissemination plan* - Committee feedback
- ▶ ACHDNC meeting on Aug 1-2, 2019
  - ▶ Draft report and dissemination plan - Committee feedback
- ▶ ACHDNC meeting on Nov 7-8, 2019
  - ▶ Charge Final report, initiate dissemination activities, and manuscript preparation
- ▶ ACHDNC meeting on Feb 13-14, 2020
  - ▶ Report on dissemination activities and manuscript submission