

# Ad-Hoc Workgroup: Interpreting Newborn Screening Results Updates

Mei Baker, MD

Chair, Ad-hoc Workgroup

Advisory Committee on Heritable Disorders in  
Newborns and Children

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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

# Report Structure

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

# Part One--Introduction

## ▶ Rationale

- ▶ Limitation of NBS may have not sufficiently appreciated
- ▶ Inconsistent terminology may inadvertently lead to confusion between “screening” and diagnosis”



Unattainable Expectations

# Part TWO—NBS and Current Practice

- ▶ Screening definition: “The systematic application of a test, or inquiry, to identify individuals at sufficient risk of a specific disorder to warrant further investigation or direct preventive action, amongst persons who have not sought medical attention on account of symptoms of the disorder.” (Gilbert, 2011)
- ▶ Additional unique NBS aspects (newborns, time critical conditions, technologies, etc.)
- ▶ Possible NBS testing outcomes
- ▶ Terminology clarification
- ▶ 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—NBS and 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
- ▶ Challenges

# Timeline

- ▶ ACHDNC meeting on Nov 7-8, 2019
  - ▶ Present draft report - Committee feedback
  - ▶ Present plan for seeking additional feedback
  
- ▶ ACHDNC meeting on Feb 13-14, 2020
  - ▶ present final report
  - ▶ present manuscript plan