

Ad-Hoc Workgroup: Interpreting Newborn Screening Results Updates

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

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Chair, Ad hoc Workgroup

March 22, 2019

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

Workgroup Charges

- ▶ **Charge 1: Newborn screening results interpretation**
 - ▶ Risk assessment
 - ▶ Terminology
 - ▶ Interpretation for all testing categories
 - ▶ Message delivery--communication
- ▶ **Charge 2: Recommendations regarding screening cutoff establishment and monitoring**
 - ▶ Consider both screening sensitivity and specificity
 - ▶ On-going evaluation

Approach to Addressing Workgroup Charges

▶ Charge 1: Approach

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

▶ Charge 2: Approach

- ▶ Draft possible recommendations - report to the Committee

Charge 1:

Work Plan Part One--Introduction

- ▶ Rationale and targeted audience
- ▶ Knowledge gap and unmet expectations
 - ▶ Screening vs. diagnosis
 - ▶ Terminology harmonization
- ▶ Screening test versus diagnostic test
 - ▶ References in literature
 - ▶ MOC4 from the Midwest Genetics Network
- ▶ Individual versus population (individual circumstance)

Charge 1:

Work Plan Part Two—Current Practice

- ▶ Risk assessment evidence
- ▶ Report categories
 - ▶ 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

Charge 1:

Work Plan Part Three—Discussion and Recommendations

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

Timelines

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