

Newborn Screening

Criteria Review Advisory Committee: Recommendations

INTRODUCTION

On March 6, 2015, the Board convened a Criteria Review Advisory Committee to: 1) review the five State Board of Health (SBOH) newborn screening (NBS) criteria to see if language is appropriate or needed to be modified, 2) determine if new criteria should be added to the five criteria, 3) identify a small set of criteria to serve as “minimum requirements” or “Qualifying Assumptions” which State Board of Health and Department of Health (DOH) staff could apply to determine if it was necessary to convene an advisory committee, and 4) develop an ethical framework or guiding principles to govern the evaluation of any new potential condition. The Criteria Review Advisory Committee made three sets of recommendations for the SBOH.

The meeting was co-chaired by Dr. Diana Yu, Board sponsor and member (SBOH) and Dr. Kathy Lofy, State Health Officer (DOH). The Criteria Review Advisory Committee comprised of members from diverse backgrounds with a stake in newborn screening. A list of the membership follows.

Donna Dorris, Senior Health Policy Analyst, OIC
Charissa Fotinos, MD, WA HCA
Peggy Harris, Save Babies Through Screening Foundation
Melissa Hughes, LM CPM, Midwife Association of WA
Neil Kaneshiro, MD, Premera Blue Cross
Lain Knowles, NBS Program, DOH
Gina Legaz, WA Chapter March of Dimes
Kathy Lofy, MD, State Health Officer, Chief Science Officer, DOH, Committee Co-Chair
Lawrence Merritt, MD, Pediatrician-Biochemical Genetics, Seattle Children’s
Susan Searles Nielsen, PhD, UW Research Assistant Professor, Department of Neurology
Thomas Paulson, MD, Medical Director, Amerigroup WA
Amy Person, MD, Health Officer, Benton-Franklin Counties Health Department
Tom Pendergrass, MD, Pediatrician, Seattle Children’s
Zosia Stanley, JD, MHA, WA Hospital Assoc-Policy Director, Access
T. Howard Stone, J.D., LL.M., C.I.P., Administrator, WA Institutional Review Board
Sheila Weiss, Genetic Counselor, UW Medical Center
Ben Wilfond, MD, Director, The Treuman Katz Center for Pediatric Bioethics; Seattle Children’s

(Two committee members were absent: Margaret Hood, MD, FAAPO, WA Chapter of AAP and Sheri Nelson, Gov’t Affairs Director, Health Care; Assoc. of WA Business.)

The project was staffed by Mike Glass and Tara Wolff (SBOH health policy advisors), Melanie Hisaw (SBOH scribe and meeting organizer), and Amanda Kimura (DOH scribe and presenter).

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COMMITTEE RECOMMENDATIONS

THREE GUIDING PRINCIPLES

Three guiding principles govern all aspects of the evaluation of a candidate condition for possible inclusion in the NBS panel.

1. Decision to add a screening test should be driven by evidence. For example, test reliability and available treatment have been scientifically evaluated, and those treatments can improve health outcomes for affected children.
2. All children who screen positive should have reasonable access to diagnostic and treatment services.
3. Benefits of screening for the disease/condition should outweigh harm to families, children and society.

QUALIFYING ASSUMPTIONS

Before an advisory committee is convened to review a candidate condition against the Board's five newborn screening requirements, a preliminary review should be done to determine whether there is sufficient scientific evidence available to apply the criteria for inclusion.

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PROPOSED REVISIONS TO THE BOARD'S FIVE CRITERIA

FOR ADDING DISORDERS TO THE NBS PROGRAM

No new criteria were added to the Board's current list of criteria. However, the following revisions to the five criteria were recommended. These revisions are intended to make the criteria clearer when they are applied.

Prevention potential and medical rationale. The term "clear benefit" in the original criterion was removed because it was judged hard to quantify. The criterion was further clarified to make explicit that while identification of the condition or disease must take place in the newborn period (which generally refers to the first 28 days of life) diagnosis and intervention can occur outside of the newborn period.

Treatment available was edited to emphasize the availability of both diagnosis and treatment services. The term "appropriate" was removed because it was perceived to be subjective and difficult to define.

Public health rationale was edited to remove the parenthetical phrase "(symptoms are usually absent, such that diagnosis is delayed and treatment effectiveness is compromised)". The committee believed that the parenthetical phrase is not clear and there are potentially other reasons that could justify using population-based newborn screening. Also, reference to the prevalence of the condition was removed and is now factored into the final analysis, cost-benefit/cost-effectiveness.

Available Technology – this title was edited to specify that it applies to screening technology

Cost-benefit/cost-effectiveness was retained as the title. The committee felt that there were often circumstance when both cost-benefit and cost effectiveness should be considered in the decision-making. However, it was clarified that the analysis is to consider "outcomes" of both the screening and the treatment of the condition to put more of an emphasis on accountability for the effect on the child.

The following presents the current and adjusted language for each of the criteria.

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ORIGINAL:

- 1) **PREVENTION POTENTIAL AND MEDICAL RATIONALE:** Identification of the condition provides a clear benefit to the newborn: preventing delay in diagnosis; developmental impairment; serious illness or death.

NEW:

PREVENTION POTENTIAL AND MEDICAL RATIONALE: The newborn identification of the condition allows early diagnosis and intervention.

Important considerations:

- There is sufficient time between birth and onset of irreversible harm to allow for diagnosis and intervention.
- The benefits of detecting and treating early onset forms of the condition (within one year of life) balance the impact of detecting late onset forms of the condition.
- Newborn screening is not appropriate for conditions that only present in adulthood.

ORIGINAL:

- 2) **DIAGNOSIS AND TREATMENT AVAILABLE:** Appropriate and effective screening, diagnosis, treatment, and systems are available for evaluation and care.

NEW:

DIAGNOSTIC TESTING AND TREATMENT AVAILABLE: Accurate diagnostic tests, medical expertise, and effective treatment are available for evaluation and care of all infants identified with the condition.

ORIGINAL:

- 3) **PUBLIC HEALTH RATIONALE:** Nature of the condition (symptoms are usually absent, such that diagnosis is delayed and treatment effectiveness is compromised) and prevalence of the condition justify population-based screening rather than risk-based screening.

NEW:

PUBLIC HEALTH RATIONALE: Nature of the condition justifies population-based screening rather than risk-based screening or other approaches.

ORIGINAL:

- 4) **AVAILABLE TECHNOLOGY:** Sensitive, specific and timely tests are available that can be adapted to mass screening.

NEW:

AVAILABLE SCREENING TECHNOLOGY: Sensitive, specific and timely tests are available that can be adapted to mass screening.

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ORIGINAL:

5) **COST -BENEFIT/COST-EFFECTIVENESS:** The benefits justify the costs of screening.

NEW:

COST -BENEFIT/COST-EFFECTIVENESS: The outcomes outweigh the costs of screening.

All outcomes, both positive and negative, need to be considered in the analysis.

Important considerations to be included in economic analyses include:

- The prevalence of the condition among newborns.
- The positive and negative predictive values of the screening and diagnostic tests.
- Variability of clinical presentation by those who have the condition.
- The impact of ambiguous results. For example the emotional and economic impact on the family and medical system.
- Adverse effects or unintended consequences of screening.

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HOW THE FRAMEWORK WILL WORK

The Three guiding principles will govern phases 1-5

WHEN	ACTION	WHO	OUTCOME
Phase one	Request review of a condition	SBOH, DOH, individual or organization	Proceed to Phase two
Phase two	Apply qualifying assumption	DOH and SBOH staff	Determine if there is sufficient scientific evidence available to apply the criteria.
Phase three	Recommendation to Board to convene Advisory Committee or not	SBOH members	Decision to move forward or not
Phase four	Apply the criteria	Advisory Committee	Use the five revised criteria
Phase five	Review Advisory Committee recommendation for inclusion or not in the NBS Program	SBOH Members	Board meeting motion passed
Phase six	Approach legislature for fee funding increase, if needed	DOH	Fee increase approved by the legislature or not
Phase seven	Change rule	DOH and SBOH staff	Revise rule
Phase eight	Implement rule	DOH or Birthing Providers	Begin screening