



# Newborn Screening Criteria Review: Proposed Changes

## Summary Report

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# Background

- ▶ 1960's Increasing interest in screening
- ▶ WHO commissioned a report on screening James Wilson (England) & Gunther Jungner (Sweden)
- ▶ 1968 – *Principles and practice of screening for disease* a public health classic. 10 criteria

# Background

## ▶ Wilson and Jungner Criteria

1. Condition sought should be an important health problem.
2. There should be an accepted treatment for patients with recognized disease.
3. Facilities for diagnosis and treatment should be available.
4. There should be a recognizable latent or early symptomatic stage.
5. There should be a suitable test or examination.
6. The test should be acceptable to the population.

# Background

- ▶ Wilson and Jungner Criteria, cont.
- 7. The natural history of the condition, including development from latent to declared disease, should be adequately understood.
- 8. There should be an agreed policy on whom to treat as patients.

# Background

- ▶ Wilson and Jungner Criteria, cont.
- 9. The cost of case finding (including diagnosis and treatment of patients diagnosed) should be economically balanced in relation to possible expenditure on medical care as a whole.
- 10. Case-finding should be a continuing process and not a “once and for all” project.

# Background

- ▶ In Washington:
  - 1967 PKU
  - 1978 CH
  - 1984 CAH
  - 1991 Hemoglobinopathies
- ▶ No specific criteria

# Background

- ▶ In Washington – 2001
  - No screening panel changes since 1991, we had fallen behind
  - SBOH in partnership with DOH convened Advisory Committee to review 9 candidate conditions
  - 5 criteria developed
    - Guided the reviews
    - Used in subsequent years to consider many new conditions

# Background

- ▶ Meanwhile – the national scene – 2000
  - Health Resources and Services Administration (HRSA) contracted with American College of Medical Genetics (ACMG) to come up with a panel of conditions that should be screened by all states.
  - Report published 2005 with 29 recommended conditions + 25 ‘secondary’ conditions
  - Secretary of HHS accepted the recommendations (Recommended Uniform Screening Panel – RUSP)

# Background

- ▶ The national scene, cont.
- ▶ Most states adopted the recommendations.
  - Washington reviewed the recommended conditions using the 5 criteria and adopted most of the ‘core panel’
- ▶ But ACMG process was highly criticized for lack of a rigorous evaluation of the evidence supporting screening.

# Background

- ▶ The national scene, cont.

## Changes made in response to criticism

In depth nomination process

Much more rigorous review/documentation

Careful reviews may lead to:

- Full evidence review by contracted university
- Recommendation to Secretary HHS for inclusion on the RUSP

# Background

- ▶ Current Issues
- ▶ Advancing technology allows us to screen for more conditions.
- ▶ Current 5 SBOH criteria have served well for 15 years but,
  - Should they be revised to be more specific?
  - Little guidance for evaluating impact of late onset, highly variable manifestations.
  - Little guidance for evaluating public health role for ‘point of care’ screening.

# Background

- ▶ Current Issues, cont.
- ▶ To address these issues:
  - SBOH passed motions this fall to:
    - Review current criteria and consider adding additional criteria
    - Consider creating an ethical framework to guide the evaluation of disorders
    - Consider assumptions to qualify conditions for consideration
- ▶ An advisory committee was convened to help and provide input to this process

# Advisory Committee

March 6, 2015

- ▶ Broad representation:
  - Parent/child advocacy
  - Pediatric specialty
  - Genetic counseling
  - Bioethics
  - Human subject research protection
  - Hospitals
  - Midwives

# Advisory Committee

March 6, 2015

- ▶ Representation cont.
  - Private insurers
  - Public payers (HCA)
  - Office of the Insurance Commissioner
  - Local public health
  - DOH newborn screening program
  
  - Co-chairs:
    - Diana Yu, MD, State Board of Health
    - Kathy Lofy, MD, State Department of Health

# Recommendations

## QUALIFYING ASSUMPTION

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.

# Recommendations

## THREE GUIDING PRINCIPLES

Three guiding principles are to govern 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.

# Recommendations

## THREE GUIDING PRINCIPLES cont.

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 children, families, and society.

# Recommendations

Criterion #1 ORIGINAL:

**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.

Criterion #1 NEW:

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

# Recommendations

## Criterion #1 NEW cont.:

### 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.

# Recommendations

Criterion 2 ORIGINAL:

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

Criterion 2 NEW:

**TREATMENT AVAILABLE:** Effective diagnosis, treatment and systems are available for evaluation and care of all infants identified with the condition.

# Recommendations

Criterion 3 ORIGINAL:

**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.

Criterion 3 NEW:

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

# Recommendations

Criterion 4 ORIGINAL:

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

Criterion 4 NEW:

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

# Recommendations

Criterion 5 ORIGINAL:

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

Criterion 5 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.

# Recommendations

## Criterion 5 NEW cont.:

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.

# Adding new conditions to Washington's Newborn Screening Panel

NOTE: The three guiding principles will govern phases 1–5

WHEN	ACTION	WHO	OUTCOME
Phase one	Request for 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

# Adding new conditions to Washington's Newborn Screening Panel cont.

NOTE: the three guiding principles apply to phase 1-5

WHEN	ACTION	WHO	OUTCOME
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

# Newborn Screening Criteria Review: Proposed Changes – Summary

Questions?