State of the States Newborn Screening: Challenges and Opportunities

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State of the States
Newborn Screening: Challenges and Opportunities

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Newborn Screening

The term is used to refer to two programs that may or may not have linkages with each other:

1. Traditional biochemical screening for inherited conditions (metabolic, endocrine, hematological, etc.)

2. Screening for congenital hearing loss
Newborn Screening

Multiple Ways of Defining:

• An **essential public health program** that prevents catastrophic health consequences through early detection, diagnosis and treatment.

• A **complex system** of testing, evaluation, and treatment that is dependent upon the dedication of persons working within the system.

• A **public health prevention strategy** that when designed properly, reaches **all** newborns rapidly and effectively.
A Public Health Prevention System that includes

- Family Members
- Primary Health Care Professionals
- Laboratory Personnel
- Administrative and Follow-up Personnel
- Specialty Care Centers
- Source(s) of Payment
- Policy Makers
- Manufacturers
- Other Interested Persons or Groups
Newborn Screening

How did it start?
Brief Review:
Newborn Screening History

1960s

- Guthrie developed filter paper test for PKU. (Identified newborns with PKU whose diet could be modified thus preventing mental retardation.)
Brief Review: Newborn Screening History

1960s

- Parents pressed for testing through organized lobbying.
- Legislatures began to pass mandatory newborn screening laws to reduce institutionalization.
- Guthrie developed other filter paper metabolic tests.
Brief Review: Newborn Screening History

1970s

- Other filter paper tests became available – Congenital Hypothyroidism, Sickle Cell Anemia, etc.

- Technology improved, allowing program expansion.

- State Legislators began asking programs to become self-supporting—fees were introduced.
Brief Review:
Newborn Screening History

1990s

- DNA tests used as second tier – Sickle Cell Disease screening; Cystic Fibrosis screening

- Application of tandem mass spectrometry (MS/MS) to allow simultaneous detection of multiple disorders.

- AAP Newborn Screening Task Force
Brief Review: Newborn Screening History

2000s

- Newborn hearing screening
- Public pressure to expand testing with MS/MS
- Emphasis on program integration (including data/information)
- Privacy concerns – residual blood spot, federal HIPAA rules (data sharing)
Brief Review: Newborn Screening History

2000s

▪ Advisory Committee on Heritable Disorders and Genetic Diseases in Newborns and Children (ACHDGDNC)—2003 chartered

▪ American College of Medical Genetics (ACMG) report to HRSA—Final report 2006

▪ “Newborn Screening Saves Lives” Act – Signed 2008; No appropriations
The Issue of Benefit
The blind men and the elephant.

Poem by John Godfrey Saxe

http://www.nature.com/ki/journal/v62/n5/fig_tab/4493262f1.html
Assumption

• Advances in technology will lead to a likely scenario in which hundreds of conditions could be screened simultaneously, at very low cost and with great accuracy
Assumption

• Increasingly effective advocacy efforts and competition from the private market, will push for a rapid and significant expansion of the public health newborn screening program
Question

• Does the fact that we can screen for a condition mean that we should screen for it?
Much of the discussion about expanded screening has focused on possible risks:

- Cost
- Available supports
- Ability to scale-up
- Equity
- Ethics
- Potential for harm
Early identification provides the opportunity to optimize the infant’s environment to maximize the potential effect of environment on phenotype.

Research has already documented the primacy of the early years and the effectiveness of early intervention.

NBS provides access to family support services and information that can have positive benefits for families and prevent costs to families and society of the “diagnostic odyssey.”

For many conditions, NBS provides earlier access to an existing program of services that families endorse as both positive and effective.

Expanding NBS is consistent with research on consumer preferences for information.

Also:

• Consumers are holding professionals accountable (duty to warn)
• NBS has other benefits to science and society
  – Determine the **true incidence** rate of a condition
  – Identify the **full range** of genotypic and phenotypic expression
  – Allow for studies of **earliest development** and onset of phenotypic features
  – Will push a research and development agenda on **new treatments**
  – As new treatments become available, NBS could provide **faster access** to families of affected individuals
1. Benefit has been and remains a core consideration for screening decisions

- Almost all historical reports reach the same general conclusion: Screening should be limited to those conditions for which there is proven benefit.
- But, almost all (95%) of the 78 conditions reviewed in the ACMG report had a treatment that could prevent at least some negative consequences for the child and all (100%) could result in at least some family and societal benefits.
In discriminating among candidate conditions for NBS, question is not whether benefit occurs, but:

- The **nature** of the benefit
- The **magnitude** of benefit
- The level of benefit relative to **costs** or possibility of harm
2. Benefit has focused on **improved physical health** for the infant via medical treatment

- Most historical reports focus on this particular benefit, although “treatment” and “benefit” are often used interchangeably.
- Early intervention could prevent mortality in most (86.2%) of the core panel conditions.
- All core panel conditions had a nutritional or other medical treatment that could prevent some negative consequences.
4. Screening for all conditions has perceived benefits for family and society, BUT:

- Although all reports acknowledge the possibility of these benefits, only two (WHO, 1969; NAS, 1975) considered these as legitimate bases for policy decisions.
- Most reports consider such benefits as additive to infant benefits rather than a stand-alone justification for screening.
5. There is no agreed-upon threshold for meaningful benefit

- What level or kind of improvement in well-being should be considered sufficient?
- How do we weigh direct benefits to the infant in relation to benefits for families or society?
- What level of quality of life would form a threshold below which screening would not occur?
- These issues are complex, value laden, and difficult to quantify
- Most reports ignore this question completely, focusing primarily on the possibility of treatment
6. Stakeholders differ in their perceptions of benefit

- **Professionals** tend to endorse a more conservative stance, insisting on improved health outcomes.
- **Parents and advocacy groups** generally take a much broader view of benefit; in fact, many would argue that information about a condition would constitute sufficient benefit, regardless of treatment potential.
- For many parents, there is **no such thing as an “untreatable condition”**.
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