New Disorders and Expanded Screening:

The future of NBS

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NEWBORN SCREENING

Disclaimers:

I do not have a crystal ball.
We are not funded/supported by industry partners.

These personal opinions are motivated by participation in national committees and workgroups.

My thoughts are motivated by the struggles, sufferings and hardships of families and patients with rare diseases.

Rare Disorders and Newborn Screening:

- > 6,000 disorders
- 80% with genetic cause
- 50% affects children

- **> 2,400 NBS disorders**

Current process is not agile enough to expand screening panels by even 10-20 disorders at once.

The current disorder inclusion/nomination/selection process:

Incomplete and does not allow input from the entire rare disease community

- Lack of broad ideation and selection “funnels”
- Biased process shaped by special interest groups: e.g. large foundation or for-profit companies
- Often excludes payors in the decision process
- Process shaped by artificial boundaries that prohibit efficiencies
- Strange and scientifically unsound selection criteria

Solutions: The current disorder inclusion/nomination/selection process:

- Broad, transparent ideation and prioritization matrix (NBSTRN3.0) with timelines and regular and standardized review
- Elimination of current nomination process
- Methods of payor inclusion, real-time benefit-cost assessments?
- Continuous assessments of NBS program costs

Lack of technology development and scalable technology solutions:

- Status quo dictated by few solution providers with FDA cleared methods and solutions
- Existing scalable technologies are not developed rapidly enough; e.g. LC-MSMS solutions; WGS based methods
- Technology development does not happen in NBS programs
- NBS programs often cannot compete for development talent

Solutions: Public private partnerships or targeted grant programs with clear objectives

- Specific methodology development initiatives with specified outcomes: analytical targets (xx analytes) with specific performance outcomes

Conflict between “Screening” and “diagnostic” or “clinically actionable” results

Increasing number of disorders = increasing second tier/diagnostic requirements

→high probability that public health programs are not optimally positioned (TAT, development, efficient infrastructure)

→ Secondary cost explosions

Solutions: Public private partnerships or regionalization of resources operating under diagnostic umbrella

- Explore other community models: e.g. Coops
- Economic partnerships between industry partners and states

NBS and health inequities

When we expand screening panels, we have to be keenly aware of introducing inequities (and secondary liabilities)

Do we fast-track ultra-rare disorders with very effective treatment modalities?

How to deal with disorders without “treatments”?

Solutions:

Encourage radical innovation
National Coordination Efforts

Shift to:
Focus on real-time fractional benefits-costs analyses affecting patients with rare diseases and the public at large