“Genomic” Newborn Screening: Programmatic Implications and ELSI Implications

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An Important Distinction?

Screening Newborns vs. Newborn Screening

- More Generalized Contexts (ex. Clinical Settings)
- State Newborn Screening Programs (Public Health, Mandated, Universal)
## Programmatic Benefits

**For current conditions on NBS**

- Refining screening validity
- Identifying genomic variation
- Identifying modifiers
- Reduction of a diagnostic odyssey

**For New Conditions**

- Ability to screen for a wider set of conditions for which typical assay screening is difficult
- Ability to screen for disease susceptibility genes
Programmatic Concerns

• Costs of Technology

• Storage and Management of Genomic Information

• Interpretation of genomic results
  – Incidental Findings, VUS’s

• Communication of Results
  – Education and Counseling
  – Need for follow-up (already a problem)

• Concerns among programs about genomics as a replacement tech.
Programmatic Concerns

- Psychosocial harms associated with uncertain or ambiguous genomic data (impact on follow up)
  - Variable risk associations/Spectrum of Disease

- Moving us further from the core goals of NBS programs
  - Eroding the trust in the NBS program (privacy, bloodspot experience)
Uses of Genomic Technology in NBS

Genomics as an Adjunct Technology

Genomics as a Replacement Technology
Genomics as an Adjunct Technology

Genomics as a mandatory secondary screen

Genomics as an optional parental choice
New HRSA Funding: **Improving the Newborn Screening System in the Genomic Era**

- R40: Maternal and Child Health Policy Analysis Program (HRSA)

- Project Goal: To identify and begin addressing the programmatic and policy challenges raised by the integration of NGGS technology into NBS programs in order to preserve the effectiveness of the NBS system and, in turn, improve health outcomes for newborns and their families.
  - Co-PI’s: Aaron Goldenberg and Beth Tarini
  - Consultant: Amy Gaviglio (Minnesota)
  - 2 year project
Specific Aims/Progress

- **Aim 1.** To identify programmatic and policy challenges posed by the integration of next generation genomic sequencing into the NBS system
  - Focus Groups, Interviews and Discussions with State programs (Program Directors, Laboratory Directors, Follow-up coordinators, Others)
  - Utilizing the 7 HRSA Regional Genomics Collaboratives
  - Data Collection is ongoing (at regional meetings, and will utilize webinars/phone interviews)

- **Aim 2.** To develop, test, and disseminate toolkits for state NBS programs that address the programmatic and policy challenges identified in Aim 1
Preliminary Data

• Impact on Program
  “The amount of unknown knowledge that you would have to try and explain to parents, never mind the workforce capacity that would be required and the time constraints and just huge. I mean it would be expanded newborn screening times...I don’t even know, a million?”

• Costs
  how do you incorporate it into that cost of newborn screening? ‘Cause like a lot of states, the money we collect from newborn screening doesn’t stay with newborn screening. It goes in State General Fund, and then you have to give money appropriated to pay for newborn screening....so the cost for the healthcare system might be less, but it still has to come out of your budget, so the cost issue is an issue for any kind of sequencing
Preliminary Data

• Genomic Information vs. Biochemical Markers
  “But my part as an intervention person, I like to have the baseline, what the challenged child had, but that becomes a biomarker for monitoring too. So the genetic molecular, you only know the diagnosis, but for monitoring, we use [the biochemical baseline] for monitoring for our intervention.”

• Public Education and Program Confusion
  “I think there does have to be a strong educational component. So we’re already ten years into expanded newborn screening in our state, and I still have positives coming to clinic that they’ve been told they have a positive PKU test”
Preliminary Data

- Impact on the success of the NBS program

  “So one of mine is I mean newborn screening is under such an incredible assault anywhere and in any way in many of our states, and I’m very concerned that once it became “genetic testing” that that would really cause problems with the public perception of ‘What is it?’”

  “Yeah, and we’re not ready for that and there are so many states now where the whole newborn screening program is put in jeopardy by that, and I’d be worried that [genome sequencing] would actually make that worse”.

And I think we should not repeat the mistakes of the past, because I remember when we expanded newborn screening with tandem mass spectrometry, everybody jumped the gun. So I think we need to be smart in the way that we should gather some information on those conditions, get an idea of when to start treatment, how to follow these patients before we start the [genomic] screening.
Future Plans/Outreach

• Finding ways to connect with U19 network

• Collaborating with other groups
  – NBSTRN
  – ACMG
  – Genetic Alliance (Baby’s First Test)
Thank You!

- Beth Tarini (University of Michigan)
- Rich Sharp (Mayo Clinic)
- HRSA Child and Maternal Health Bureau (R40MC268050102)