“Genomic” Newborn Screening: Programmatic Implications and ELSI Implications

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-Time 12/13/12
An Important Distinction?

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

Screening Newborns VS. Newborn Screening
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 State NBS programs.
  - Co-PI’s: Aaron Goldenberg and Beth Tarini (Consultant: Amy Gaviglio)
  - Focus Groups, Interviews and Discussions with State programs
  - Working with the 7 HRSA Regional Genomics Collaboratives
A Spectrum of Sequencing in NBS

- **Sequencing Single Genes or Specific Genetic Variants**
- **Sequencing the Entire States Panel**
- **State Panel + known modifiers or more directed panels**
- **Whole Exome Sequencing**
- **Whole Genome Sequencing**

Programmatic Complexity

ELSI Complexity
Programmatic
Benefits vs. Concerns

- Refining screening validity
- Identifying genomic variation and potential modifiers
- Ability to screen for a wider set of conditions for which typical assay screening is difficult
- Ability to screen for disease susceptibility genes

- Costs and Data Management
- Workforce needs
- Interpretation and Communication of Results
- Psychosocial harms associated with uncertain or ambiguous genomic data
- Moving us further from the core goals of NBS programs
Education/Counseling for Parents and Providers

• “the doctor has to understand the results, and then the patient has to be taught how to understand the results...The general pediatrician may not know what to do or how to describe it, and then once they give the wrong message, it gets perpetuated incorrectly in the patient population”

• “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 saying that they’ve been told they have a positive PKU test”
Workforce Needs and Deciding What to Return

• “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?”

• “we just don't have the manpower to provide information to providers and counsel the families.”

• “If research changes over time, whose responsibility would it be to re-contact those families, to tell them, oh by the way this information has changed. We just don't have the workforce that could do that.”
Incidental Findings

• “Ethically, I think most programs feel that they need to report what they find, and as a labratorian, you report what you find. To window something out means to me that you may be missing something that might be a very key piece of information for a family. And how do you live with that.”

• “That reminds me of the Tandam Mass Spec issues, when we had incidental findings, and then Maine got sued, now everyone is saying, well now I guess we have to give parents that information”
Concerns about Scope

• “When you do population based screening, if you go looking, you will find, things you never thought you were going to find...this one makes my knees shake, with just how broad this is and the types of things we could start uncovering.”

• “Even now in diagnostic testing they don't know what mutations are disease causing or clinically significant, so we don't know what to do with these kids. So they are just followed, and I can't imagine how this would increase that exponentially.”
Concerns about Complexity

• “Even now in diagnostic testing they don't know what mutations are disease causing or clinically significant, so we don't know what to do with these kids. So they are just followed, and I can't imagine how this would increase that exponentially.”

• “[The information] is more complex...there’s gonna be more information that we need to be able to explain to the parents, and so as far as how to get it going with parents, I agree that the brochure isn’t going to cover it, and who’s going do the discussion with the parents about this, the whole-genome sequencing and what it means for them?”
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?’”

• “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.”
Universality of Newborn Screening

• “it’s one of the only places in life where there’s not healthcare [disparities]... that’s our mantra, right, is universal health? The only time in your life you really could get it, and so where can we fit in there to benefit our population? And a lot of people are very interested in using it; it’s just trying to figure out how Public Health fits in.”

• “to echo what <Participant> said just a minute ago, I think the universality is really a baseline value that I think most Newborn Screening people share”
Uses of Genomic Technology in NBS

Genomics as an Adjunct Technology

Genomics as a Replacement Technology
Genomics as an Adjunct Technology

Genomics as an optional parental choice
Genomics as a mandatory secondary screen
Lessons from Newborn Screening

Kim S et al. Pediatrics 2003;111:e120-e126
Future Plans/Outreach

• Finding ways to connect with U19 network

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

- Beth Tarini (University of Michigan)
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