

“Genomic” Newborn Screening:

Ethical Hazards, Programmatic Challenges, and Parental Interest

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Parental Interest?

- Are parents interested in WGS if it were offered through the states NBS program?
 - Specifically looking for baseline interest w/o education or consent process
- C.S. Mott Children's Hospital National Poll on Children's Health (University of Michigan)
- Nationally representative panel through Knowledge Networks



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Sample

- 1539 parents (having a child at home <18)
- Demographics:
 - 63% White (non-hispanic)
 - 56% F
 - 62% some college
- 21% planning on having another child within next 5 years



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Introduction

- *Staying healthy and getting sick are affected by many things. Our genes – which we inherit from our parents – can affect our health and illness in many ways.*
- *Genes are made of DNA, and contain the instructions needed for our bodies to grow and function. All of the genes in a person make up that person's genome.*

It is possible to study a person's entire genome. This process is called whole genome sequencing. It may give information about a person's risk of having different diseases in the future.



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NBS Scenario

- *Imagine that you have a newborn baby. Shortly after birth, your child had blood collected to test for serious diseases that affect infants. These tests are done as part of each state's newborn screening program.*

Your state newborn screening program now offers you the chance to get your child's whole genome sequenced as part of the program. You would receive the results and would not have to pay for the testing. You can decide whether or not you want the information to be a part of your child's medical record.



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Interest in WGS in NBS

- *How interested would you be in getting your newborn baby's whole genome sequenced?*
 - 36% were *definitely interested*
 - 38.3% were *somewhat interested*
 - 18% were *not interested*
 - 8% were *definitely not interested*.
- Participants more likely to be interested in WGS through NBS:
 - female (OR=1.77)
 - Parents with at least some college education (OR=2.04)
 - Parents planning on having a child in the next five years (OR=2.22)
 - Parents whose youngest child has ≥ 2 health conditions (OR=2.63)



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Factors Influencing Interest

- Participants with higher levels of interest:
 - accuracy of the test” (74% Very Important)
 - The potential for “preventing or decreasing a child’s chances of developing disease” (67%)
- Participants with lower levels of interest:
 - privacy of the results
 - “potential for results to be used to discriminate against their child”



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Concerns from Participants

- “About genome testing, I don't like it because it can reveal diseases you might be predisposed to develop, yet for so many of these conditions, we have no way of curing them, so what good is it to worry about something that you MIGHT or MIGHT NOT get? No thank you.”
- “I think whole genome sequence is wrong you can't be sure that you are going to develop these diseases or disorders it's just something else for hypochondriacs to worry about”
- “Genome testing has many implications. As most technologies, it can be used for great purposes in preventing or warding off predisposed diseases. Yet it could also be used by unscrupulous insurance companies to deny people insurance based upon what they might be predisposed to”



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Interest in WGS in NBS

- How interested would you be in getting your newborn baby's whole genome sequenced?
 - 36% definitely interested
 - 38.3% somewhat interested
 - 18% not interested
 - 8% definitely not interested.

- How interested would you be in getting your newborn baby's whole genome sequenced (w/ research)?
 - 31% definitely interested
 - 35% somewhat interested
 - 22% not interested
 - 8% definitely not interested
 - 11% definitely not interested.

*Sta. Stig. at $P < .01$



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Limitations/Future Research

- Hypothetical Cases
- Parents not given direct comparisons
- Small amount of info given in introduction
- Hard to assess motivations.....need for more research



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Discussion/Conclusions

- High level of parental interest
- May help health department/others prepare
 - Needs for education and counseling
 - Addressing particular concerns
- Overall need to be careful about what these kinds of data tell us (ex. Need for better ed on genomics)
- Need for more research (U19's, other)



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Patient Concerns

- Questions about how to incorporate this data into health decisions?
- How to manage so many results?
- Psychosocial harms associated with uncertain or ambiguous genomic data
- Parents rights to genomic information vs. “child's rights to an open future”



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Programmatic Concerns

- Storage and Management of Genomic Information
- Interpretation of genomic results
- Communication of Results
 - Education and Counseling
 - Need for follow-up (already a problem)
 - What would states be responsible for?
- Concerns among NBS programs about genomics as a replacement technology



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Programmatic Concerns cont.

- Moving us further from the core goals of NBS programs
 - Already happening with Expanded Screening MS/MS?
- Eroding the trust in the NBS program
 - Privacy of Genomic Data (Access issues)
 - Bloodspot Experience



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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



Thank You!

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