Sequencing Newborns
A Call for Nuanced Use of Genomic Technologies

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Presented to the Advisory Committee on Heritable Disorders in Newborns and Children
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Barbara Koenig accepted travel funds from Illumina to attend a policy discussion about sequencing.

Josephine Johnston has no financial conflicts of interest.
UCSF ELSI Aim 4:

- To create, in collaboration with other NSIGHT investigators and the Hastings Center, a national policy board that will develop (and disseminate) recommendations about the appropriate use of whole genome analysis in newborns.
- An example of “embedded ethics”
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<tr>
<td>Barbara A. Koenig</td>
<td>John D. Lantos</td>
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<td>Cinnamon S. Bloss</td>
<td>Megan A. Lewis</td>
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<td>Aaron Goldenberg</td>
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<td>Rachel Grob</td>
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<td>Julie Harris-Wai</td>
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<td>Josephine Johnston</td>
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<td>Galen Joseph</td>
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<td>Eric Juengst</td>
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<td>Jaime S. King</td>
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<td>Plus, invited guests</td>
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NSIGHT EAPB meetings
- November 2015 at Hastings Center
- June 2016 at Hastings Center
- February 2017 at UCSF

Workshopping of Draft Analysis and Recommendations
- June 2017 at ELSI Congress

Publication of Analysis and Recommendations plus 12 essays
- July/August 2018 special report of Hastings Center Report
Guiding Questions

- Which contextual forces shape our discussion of the utility of sequencing in newborns?
- Under what circumstances should newborns be sequenced?
- How should state-mandated newborn screening programs use sequencing?
- What role should parents play in determining how sequencing information about their infant is used and stored?
- Should sequencing be part of routine pediatric practice?
Project’s Findings

Josephine Johnston
The Hastings Center
The Ethics of Sequencing Newborns
Reflections and Recommendations

Lead article by:
Josephine Johnston
John D. Lantos
Aaron Goldenberg
Flavia Chen
Erik Parens
Barbara A. Koenig &
members of the NSIGHT Ethics and Policy Advisory Board

Plus: 12 essays by members of the NSIGHT Ethics and Policy Advisory Board
The Ethics of Sequencing Newborns
Reflections and Recommendations

A Call for Nuanced Use of Genomic Technologies
2 Reasons

- Diagnosis
- Screening

2 Types of Sequencing

- Targeted
- Whole-exome or whole-genome

3 Contexts

- Clinical Contexts
  - Sick newborns, e.g. in NICU
  - Routine primary care

- Public Health
  - In the US, state newborn screening programs

- Direct-to-Consumer
  - E.g. BabyGenes, 23andMe
Recommendation

Clinical Contexts

☐ Use targeted or whole-genome sequencing for diagnosis
  ▪ With parental permission, genetic counseling, follow-up care
  ▪ Return results that may benefit infant and/or family members

☐ Do not use as a screening tool
  ▪ Limited usefulness in asymptomatic infants
  ▪ Concerns over storage of results
  ▪ Concerns over discrimination or insurance uses
  ▪ Potential for results to generate unnecessary distress
  ▪ Potential for results to require counseling and generate unneeded follow-up care and monitoring
Recommendation

Public Health Context

- **Do not use targeted or whole-genome sequencing as sole screen**
  - Cannot detect everything
  - Concerns over storage of results
  - Concerns over discrimination or insurance uses
  - Potential for results to generate unnecessary distress
  - Potential for results to require counseling and generate unneeded follow-up care and monitoring

- **OK to use targeted sequencing**
  - As a secondary test following a positive screen
  - As a primary screen to detect conditions that meet all screening criteria
**Recommendation**

**Direct-to-Consumer**

-Parents should not use DTC sequencing for diagnosis or screening
-Health care professionals should recommend against DTC use of sequencing in infants and children
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