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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Conflicts of Interest

- Barbara Koenig accepted travel funds from Illumina to attend a policy discussion about sequencing
- Josephine Johnston has no financial conflicts of interest

Introduction: About the Project

Barbara A. Koenig, PhD

University of California, San Francisco

- 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”

NSIGHT Ethics and Policy Advisory Board Membership

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Stacey Pereira
Cynthia M. Powell
Jennifer M. Puck
Scott Roberts
Monica E. Smith
Stefan Timmermans
Rachel L. Zacharias

Plus, invited guests

Project Timeline

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

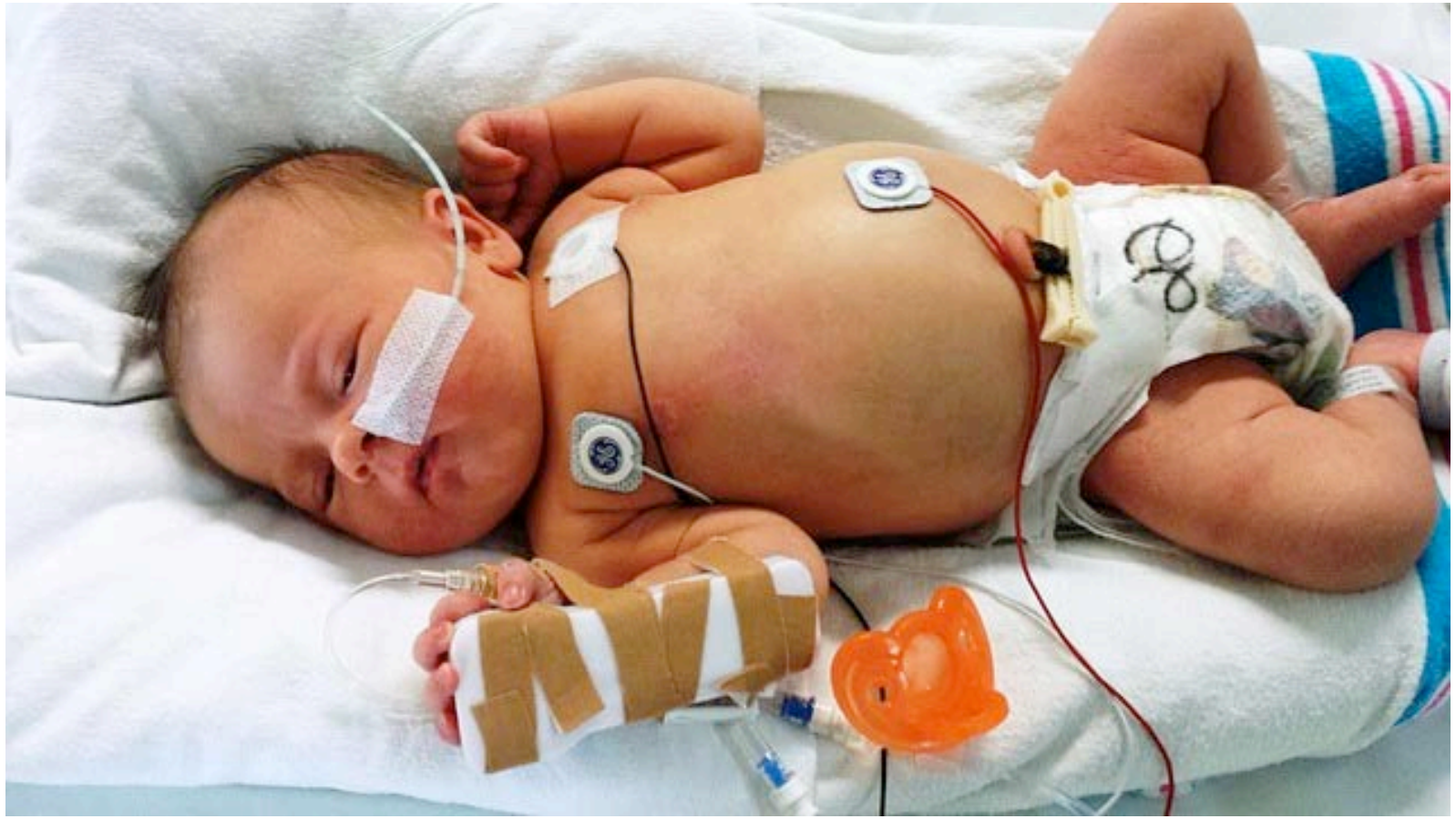
Publication of Analysis and Recommendations plus 12 essays

- ❑July/August 2018 special report of *Hastings Center Report*

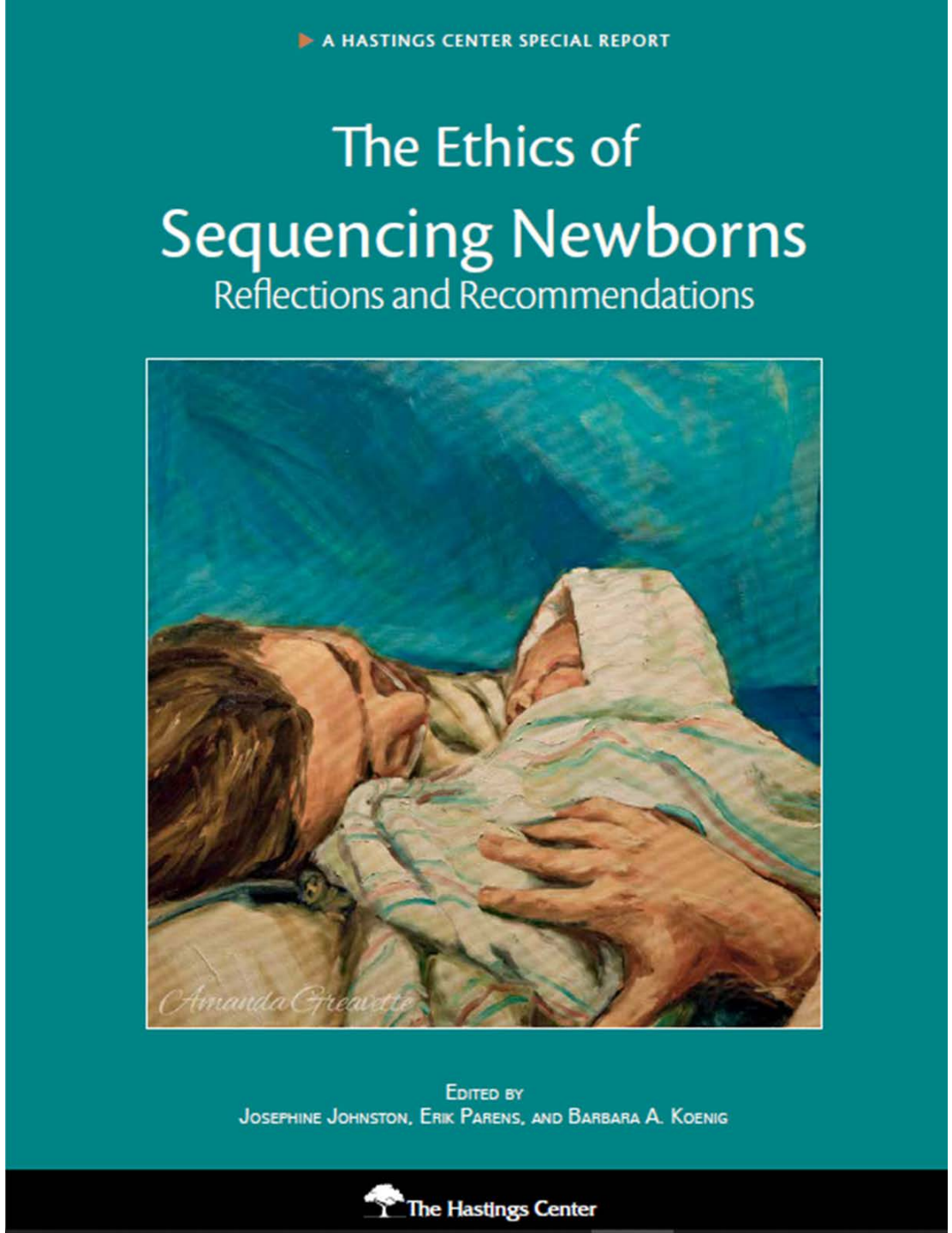
Guiding Questions

- ❑ 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

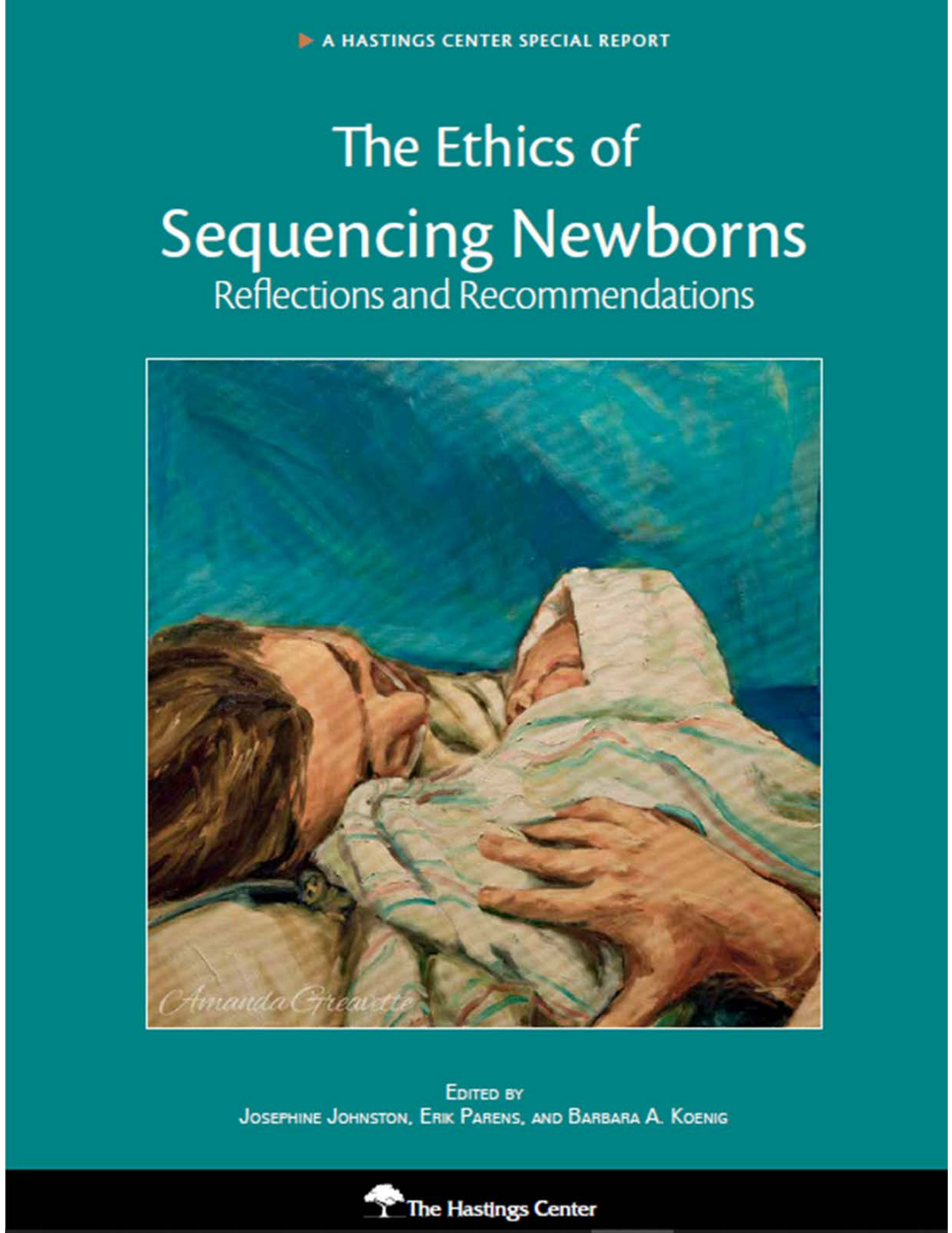


Josephine Johnston
The Hastings Center



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



Lead article

Sequencing Newborns

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Analysis

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

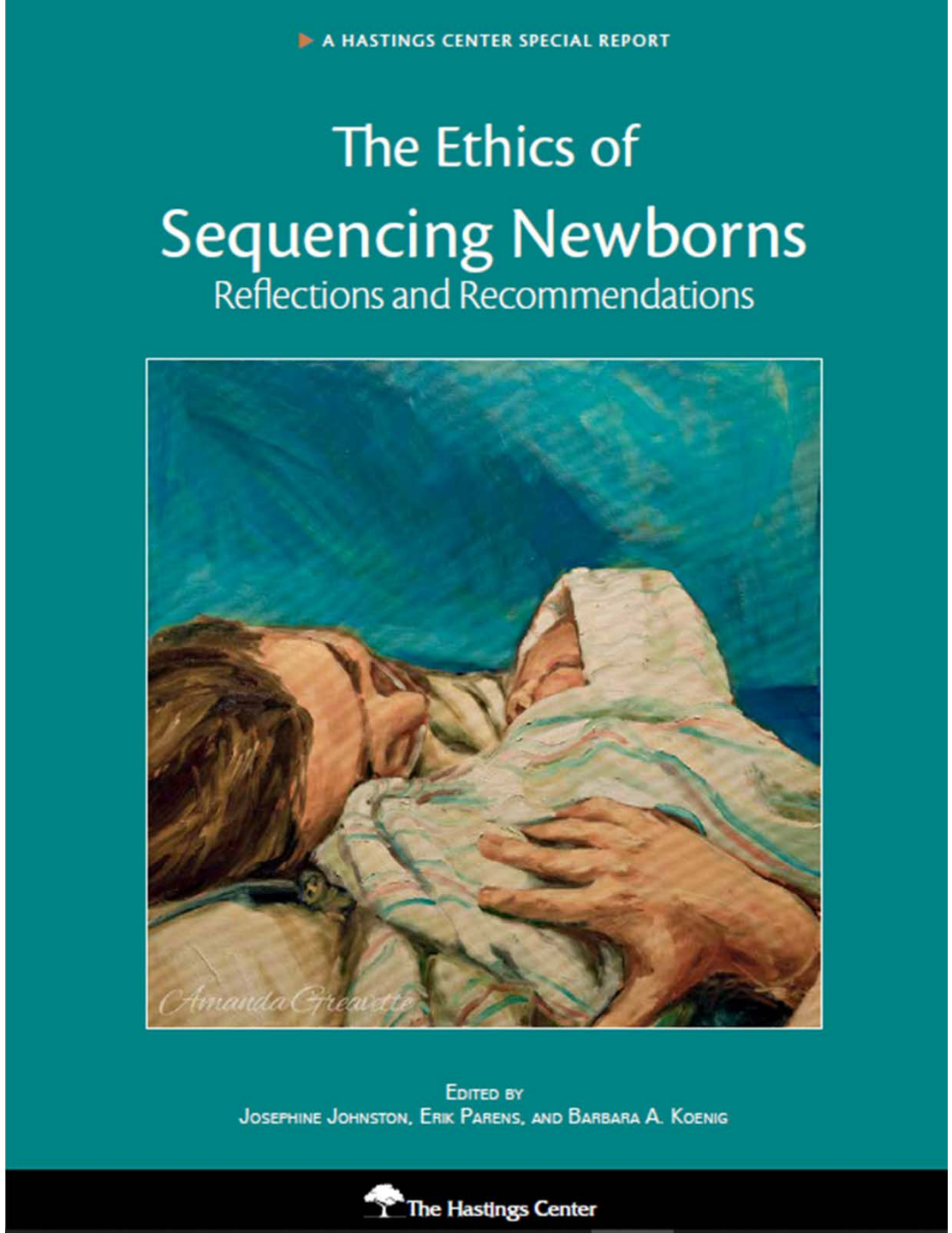
Public Health Context

- ❑ 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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