



Global Leaders in Genomic Medicine

Challenges in the Genomic Sequencing and Newborn Screening Disorders Program

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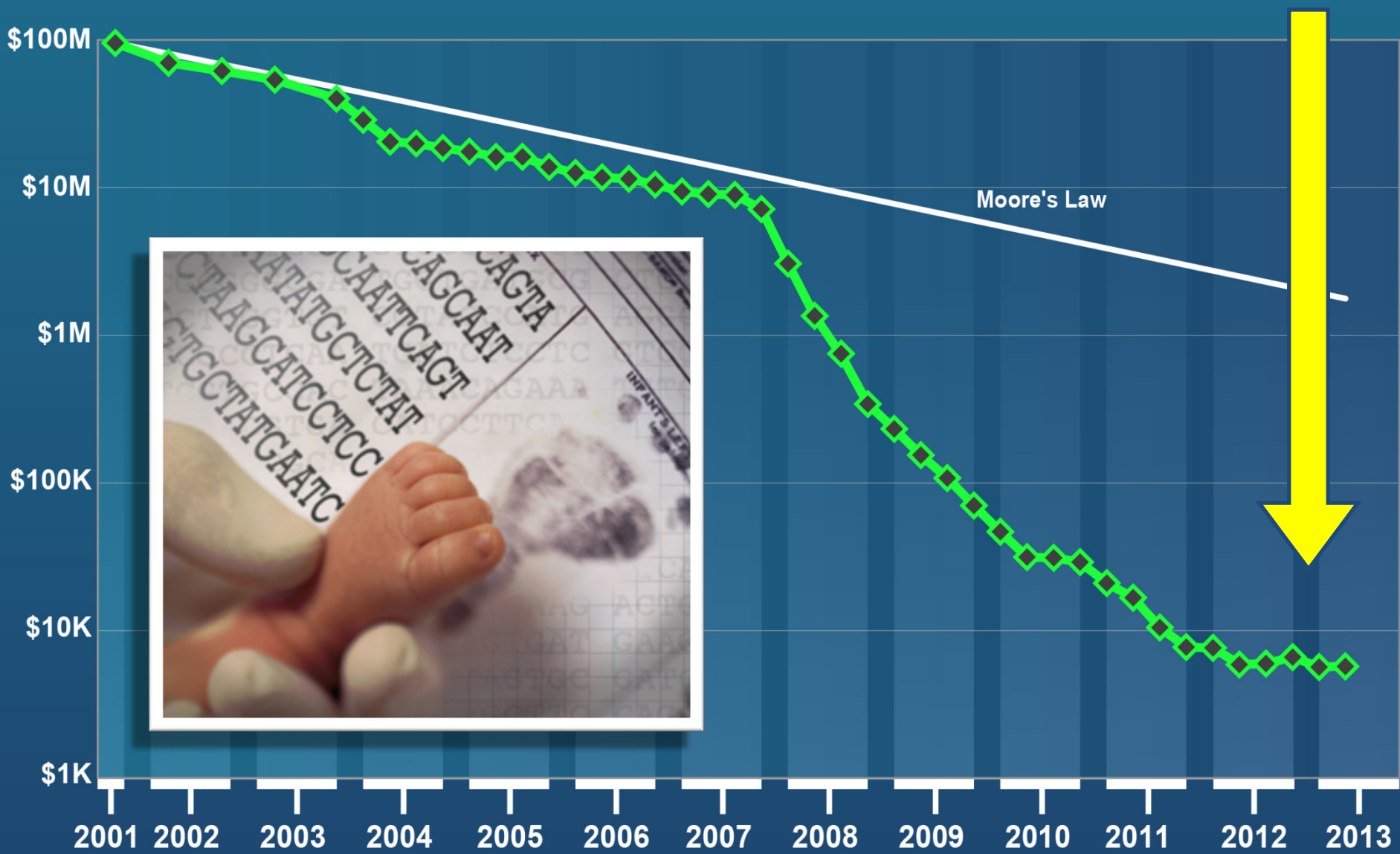
National Human Genome Research Institute



Eunice Kennedy Shriver National Institute
of Child Health and Human Development



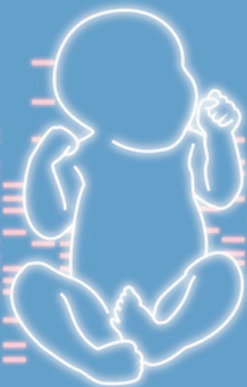
Cost per Genome





NBS in the Genomic Era

NEWBORN SCREENING IN THE GENOMIC ERA: SETTING A RESEARCH AGENDA



5635 Fishers Lane, Rockville, MD
December 13–14, 2010

SPONSORED BY:

Eunice Kennedy Shriver National Institute of Child Health and Human Development (NICHD)
National Human Genome Research Institute (NHGRI)
NIH Office of Rare Diseases Research (ORDR)



National Human
Genome Research
Institute



- Examine what genomic sequencing adds to newborn screening (NBS)
- Move towards integrating genomics in health care, anticipate challenges
- Explore ethical, legal, and social implications
- Address clinical validity and utility
- Focus on higher-risk individuals
- Incorporate longitudinal data



Advisor Comments

- ELSI concerns over population selection and study design
- Concern over public perception, public relations, and consent



NIH Purpose

To explore, in a limited but deliberate manner, opportunities to use genomic information for broadening our understanding of diseases identified in the newborn period.





Research Questions

Must address one or more of the following:

A.

For disorders currently screened for in newborns, how can genomic sequencing replicate or augment known newborn screening results?

B.

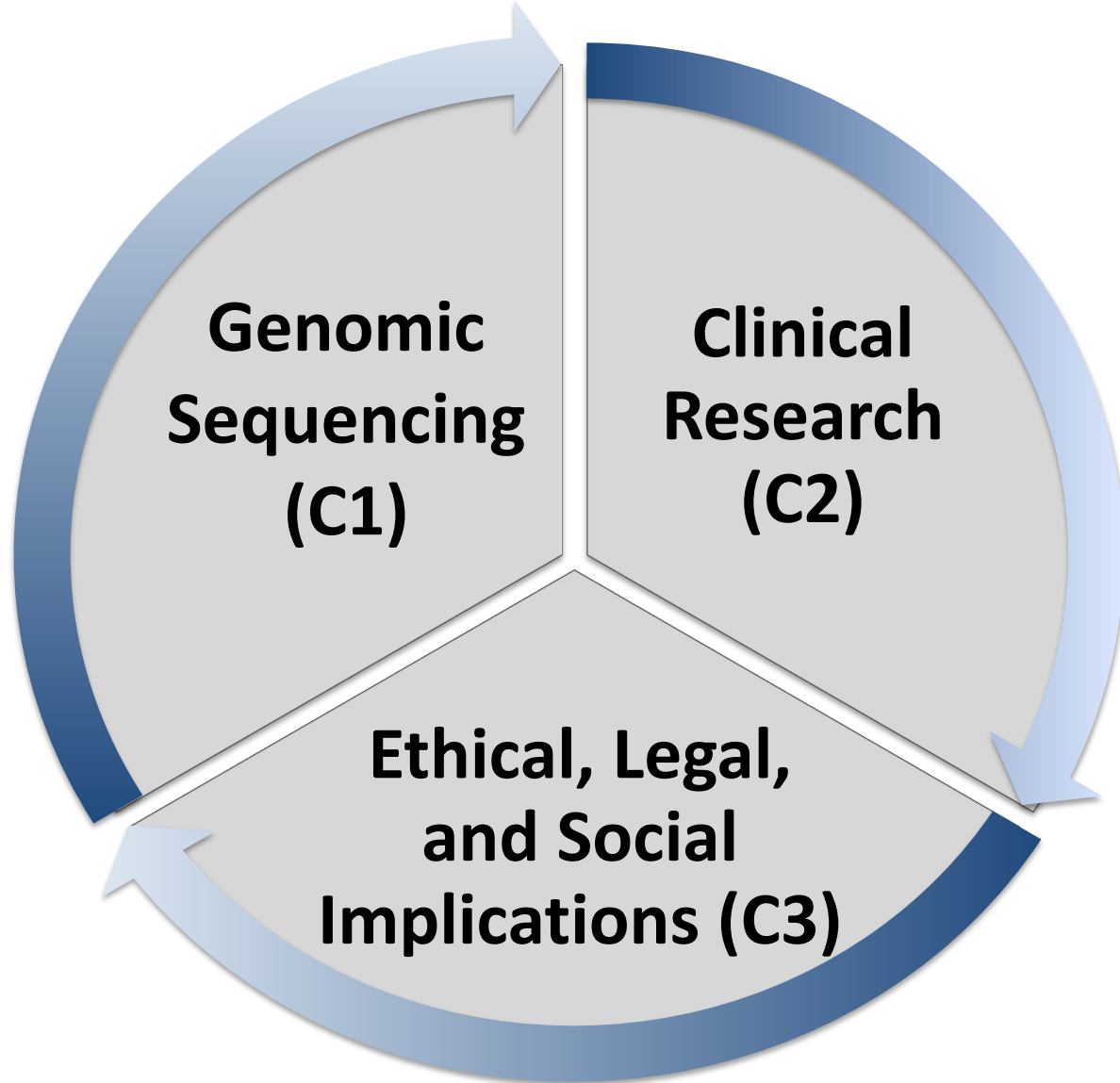
What knowledge about conditions not currently screened for in newborns could genomic sequencing of newborns provide?

C.

What additional clinical information could be learned from genomic sequencing relevant to the clinical care of newborns?



Required 3 Components






Tele-briefing and Press Release

genome.gov
National
National I

Research Funding Resea

Home > Newsroom > Current New
Telebriefing

Genomic Sequenci



Good medicine or TMI? What happens if you screen all of a baby's genes?

Maggie Fox , NBC News

Sep. 4, 2013 at 5:29 PM ET

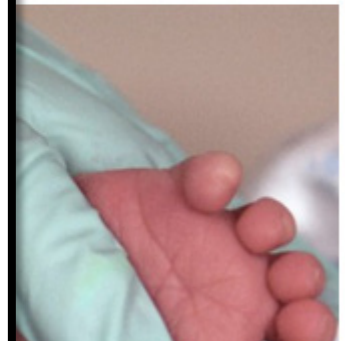
The U.S. government is funding an ambitious set of studies into what happens if you screen the entire genome of newborns. The goal is to find out whether such testing brings better health care or unwanted information overload.



SEARCH

Policy

of Sequencing



THE WALL ST

U.S. NEWS

Researchers Study Value of DNA Analysis in Screening Newborns

Initiative to determine whether sequencing can improve on "heel stick" blood test



Questions from FDA

- Need for Investigational Device Exemptions for next generation sequencers
- Thinking about device not process (CLIA/CAP)
- Concern over sequence data being returned for clinical use or placed in EMRs





Acknowledgments

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Any Questions?

THANKS!