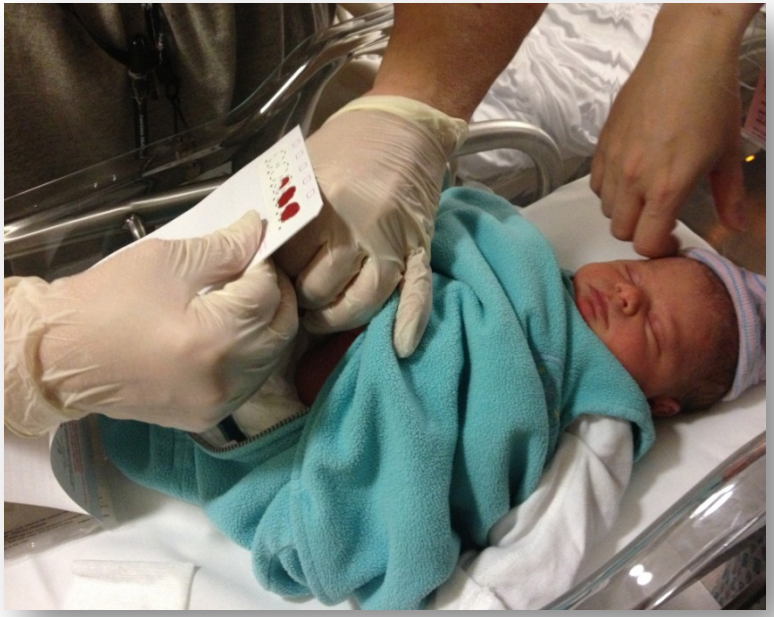
UPDATE 2015:

NICHD/NHGRI Newborn Sequencing Program

Newborn **S**equencing **I**n **G**enomic medicine and public **H**eal**T**h (NSIGHT)



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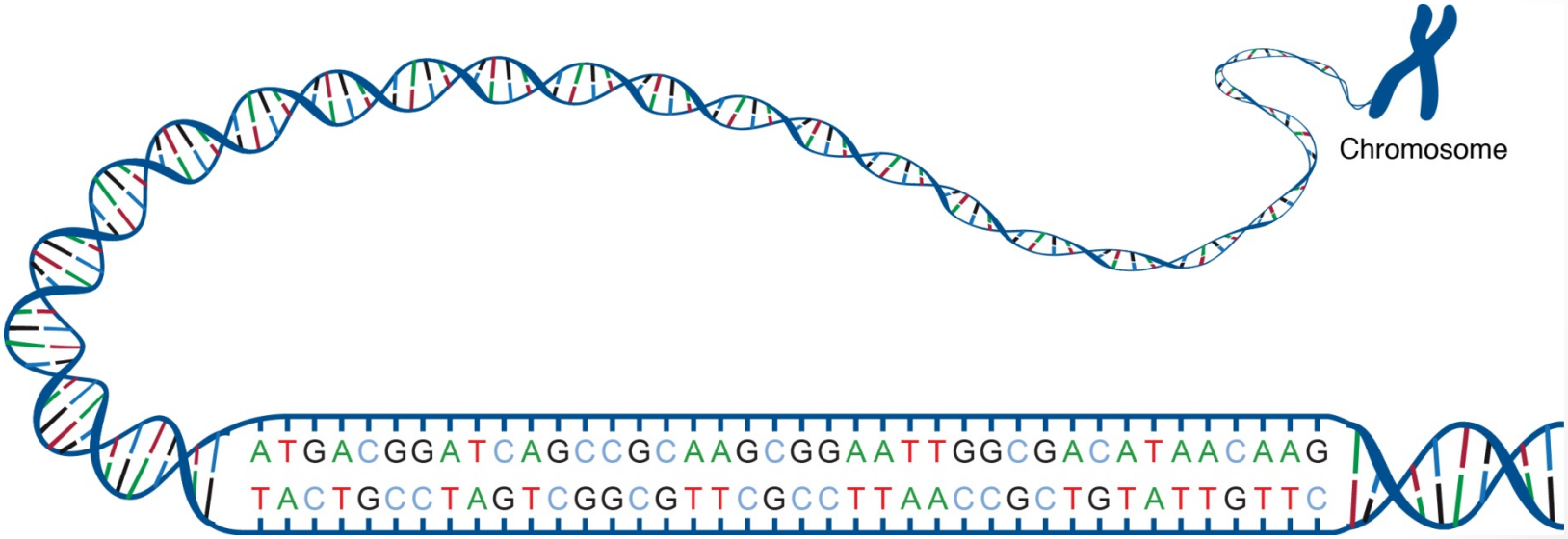
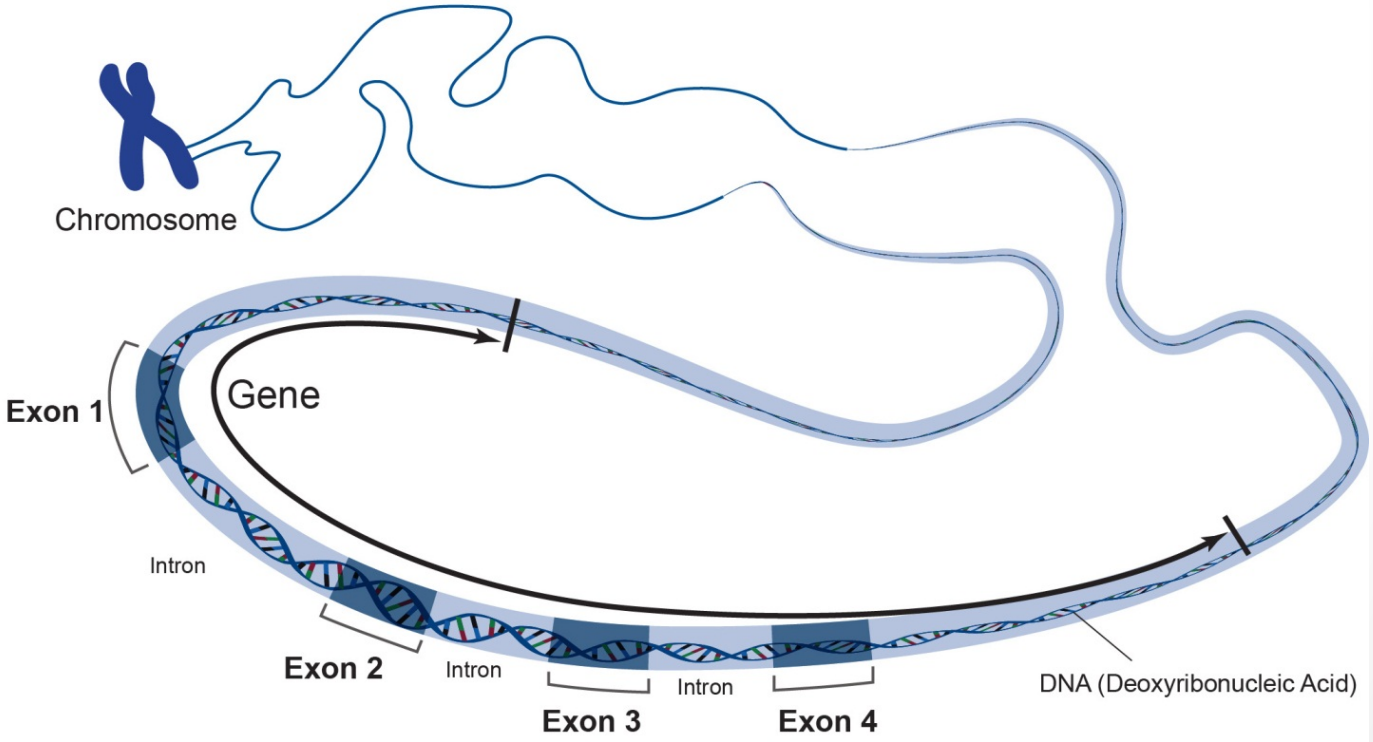
Introduction to DNA Sequencing

DNA Sequencing determines the order of the four chemical building blocks (A, C, G, T) called "bases" that make up the DNA molecule.



Introduction to DNA Sequencing

Sequence information can be used to determine which stretches of DNA contain genes and which stretches carry regulatory instructions, turning genes on or off.



Introduction to DNA Sequencing

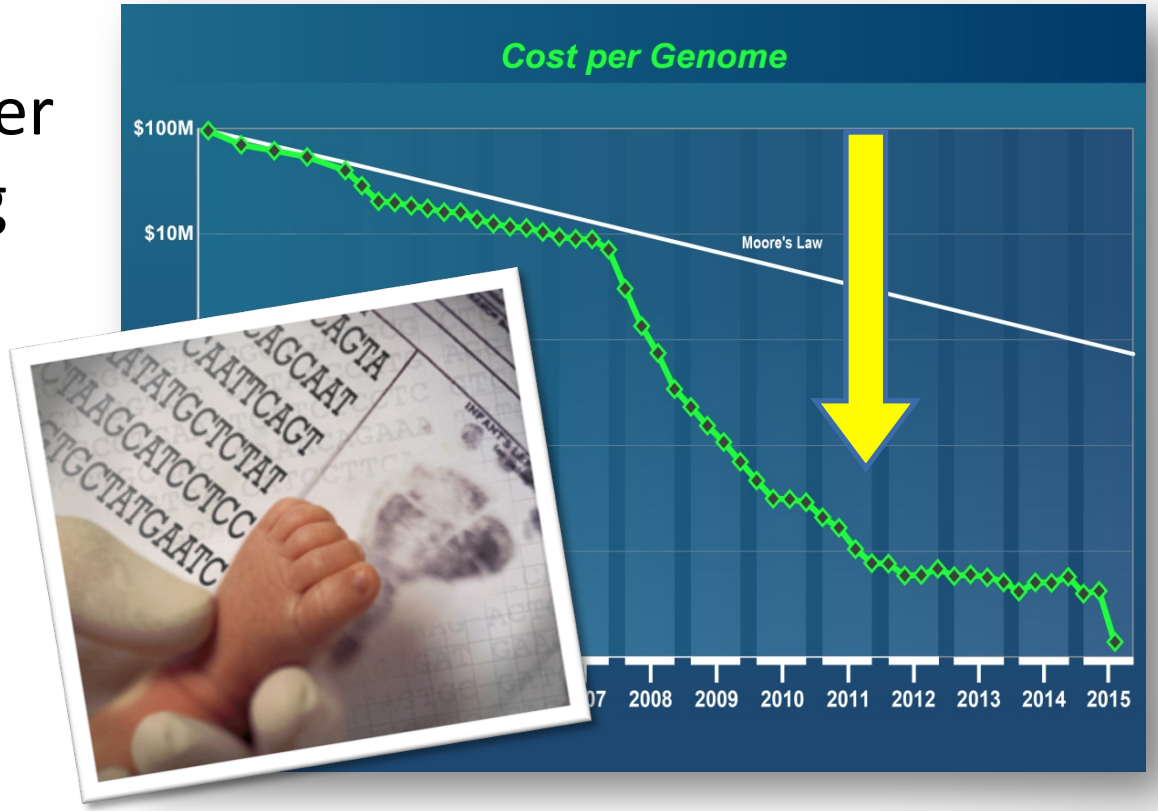
- **Exome Sequencing** selectively sequences the exons or coding regions of the genome, about 1% of the human genome.
- **Genome Sequencing** sequences most of the genome at once, over 3 billion base pairs in humans.

Genome

Exome = 1%

Genomic Sequencing

- Cost Decreasing
- Development of new and faster sequencing technologies
- Gaining a better understanding of variation in the human genome



Newborn Screening

- Highly successful national public health program
- Screening ~4 million babies a year in the United States
- Identifies babies with serious conditions so that treatment can begin before harmful effects happen

What If…

Genomic Sequencing & Newborn Screening Disorders

- NIH coordinated a meeting 2010
- Experts from academia, industry, and federal agencies in the fields of newborn screening and genomics participated.
- Outcomes
  - Important to evaluate genomic data in newborns using newborn screening as a framework
  - Important prioritize clinical validity and clinical utility; not just analytical validity
  - Important to address ethical, legal and social concerns

Research Focus

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

Goal to Examine 3 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?

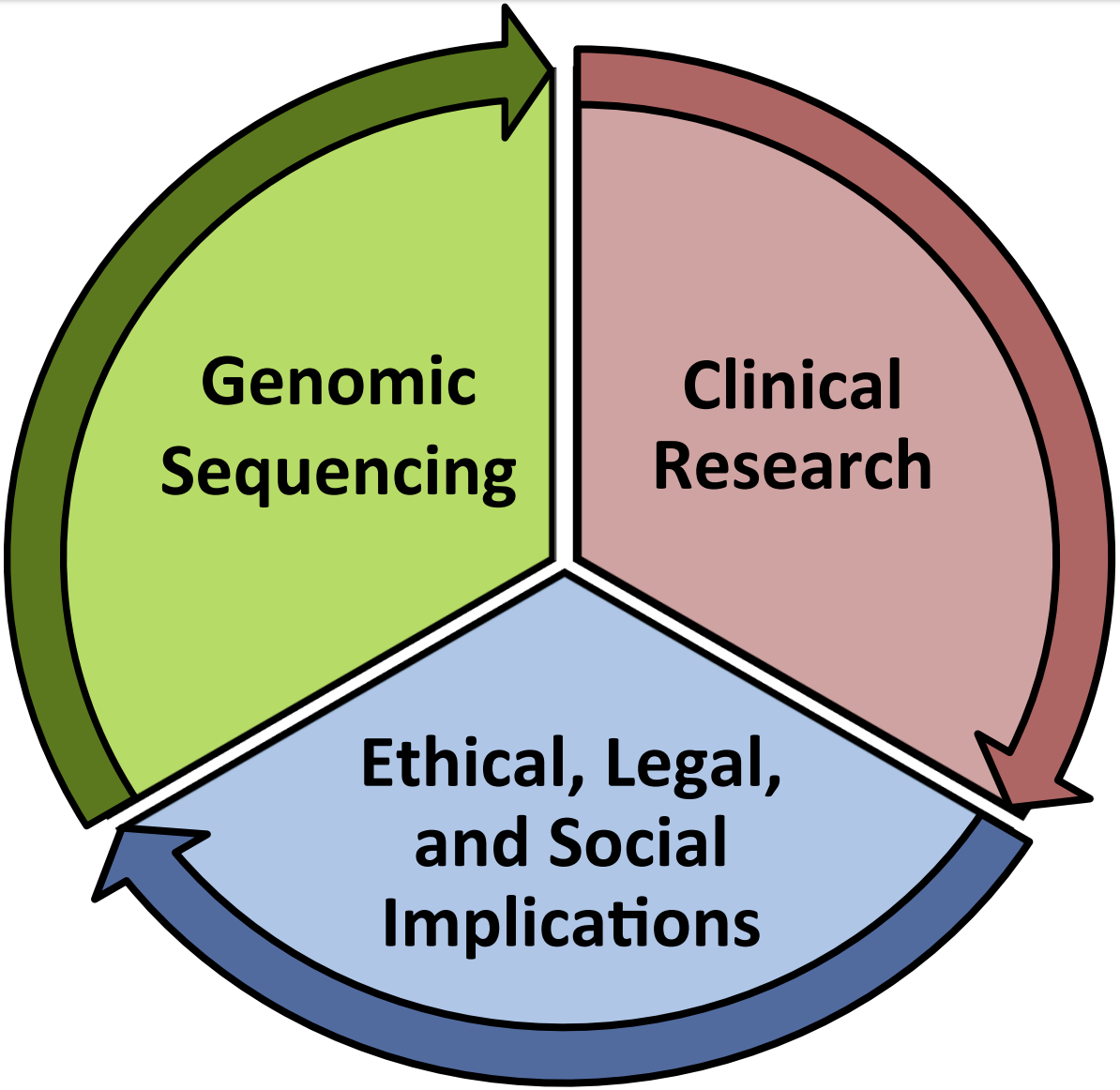
B.

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



Awardees of the NSIGHT Grants

- **Robert Green, M.D., and Alan Beggs, Ph.D.**
  Brigham and Women's Hospital, Boston
- **Stephen Kingsmore, M.D.**
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- **Cynthia Powell, M.D., M.S., and Jonathan Berg, M.D., Ph.D.**
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