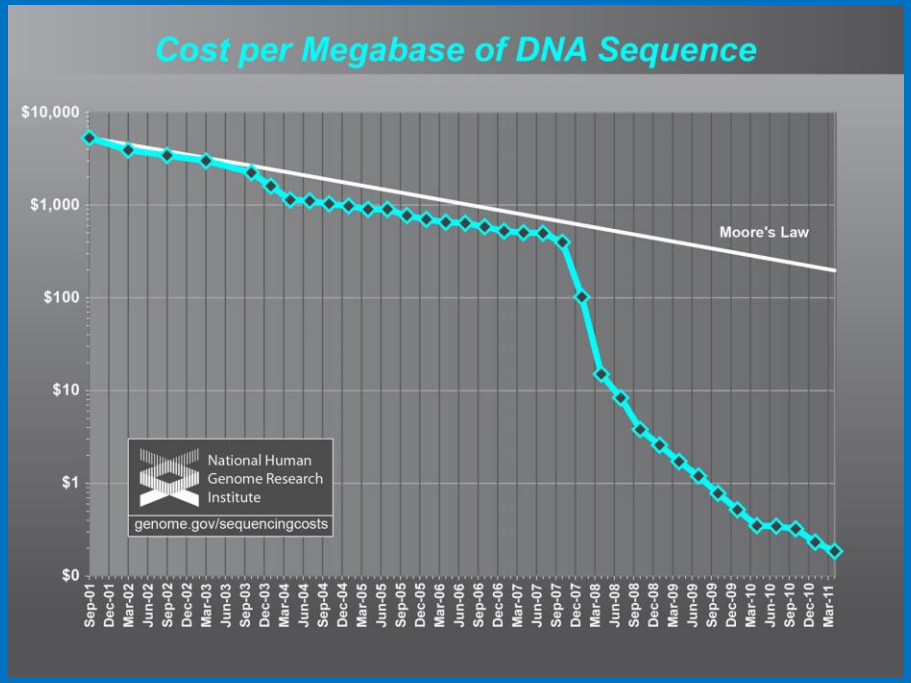
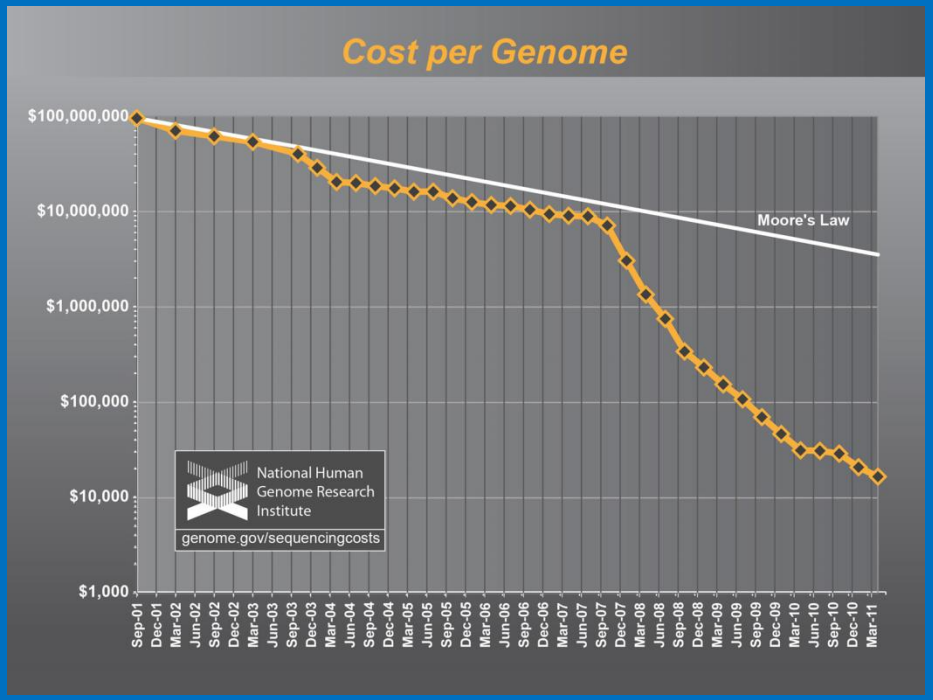
NHGRI Sequencing Program: Recent History, and Future Directions

NACHGR September 12-13, 2011

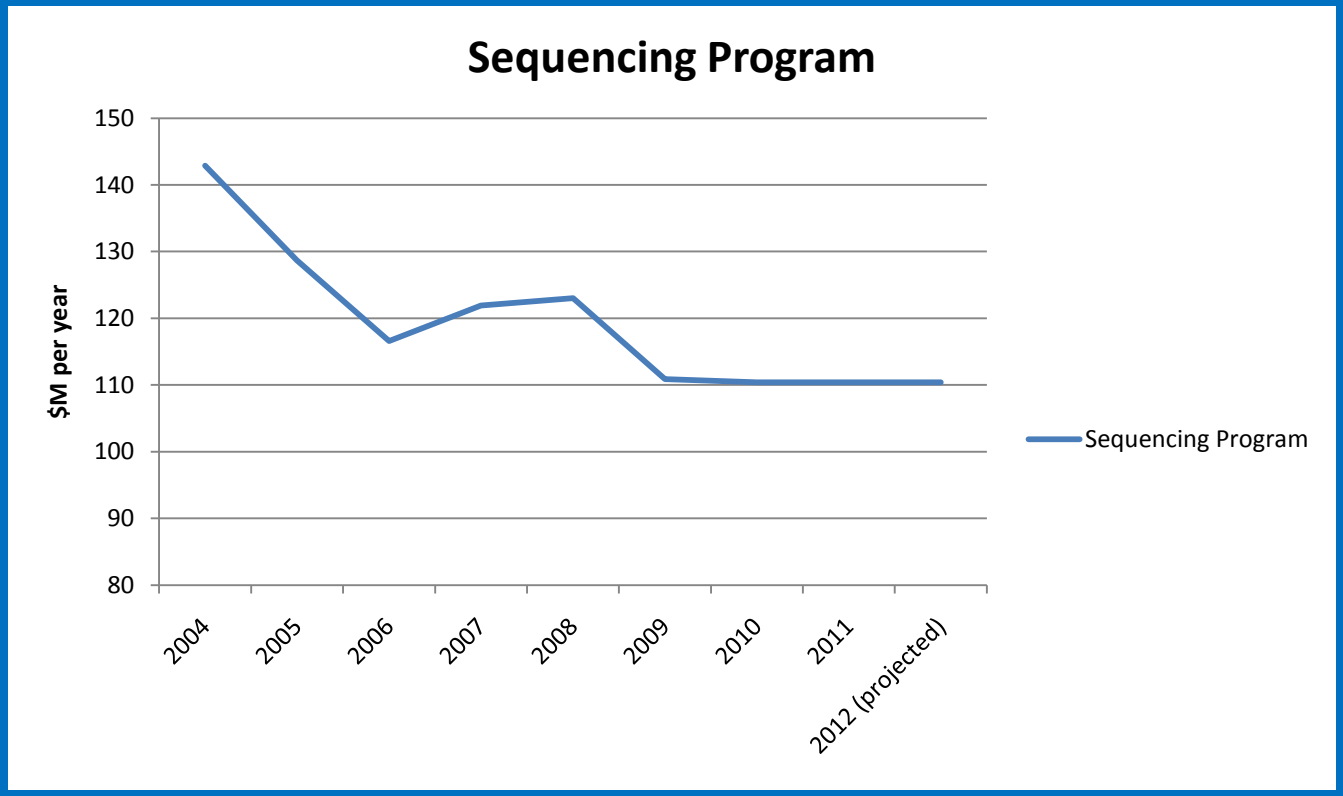
Recent History: Decreasing Cost…



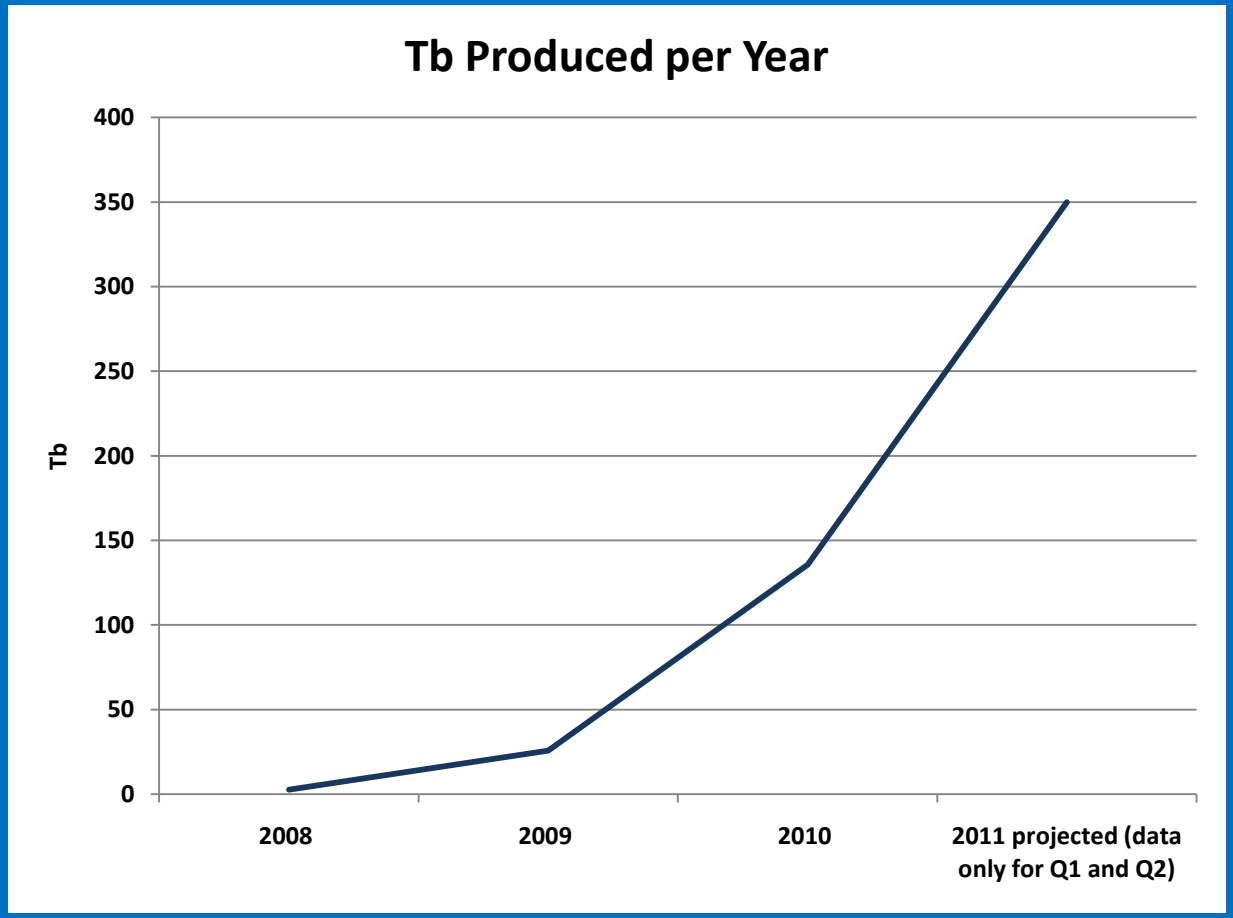


Allows Reduction of Funding Over Time…

Funding History ($M/Year)



But Still Increasing Capacity.



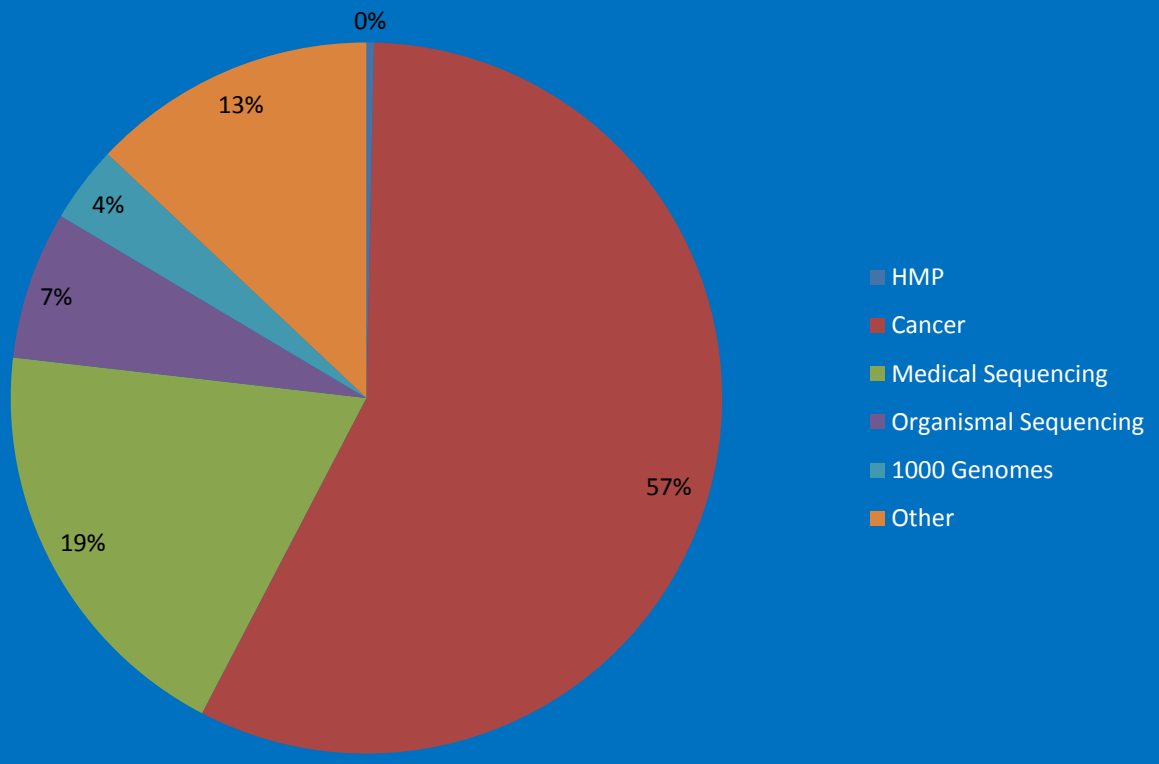
(350 Tb is equivalent to ~3500 whole genomes @ 30X coverage)

Accomplishments of the Program in Last 5 Years

Many new project types:

- Cancer sequencing
- Complex disease studies- diabetes, autism, etc.
- Mendelian diseases
- Variation resources (1000 G)
- Many organisms (comparative genomics, pathogens and vectors, models for disease); increasing inclusion of RNAseq
- Metagenomics
- *Generally building the knowledgebase for interpreting human variation*

Current Project Breakdown (Year 8 Q2)



Benefits of the Program

- Building community resources of inestimable value
- Disseminating tools and technical “know-how”
- Promoting a data sharing ethic for genomics
- Leading standards for formats and quality
- Creating templates for project design
- Pioneering new project types (e.g, whole exome capture); entails developing new methods and adapting them to new platforms; entails new analysis methods

Benefits of the Program

- Improving and maintaining the reference sequence
- Finishing/refinement: Maintaining commitment to, and capacity for, very high quality data when needed; developing new platforms with an eye to the need for this quality (structural variation, organisms)

After Major Gains, Where Next?

Context: Strategic Plan for Genomics

Context: LS Sequencing Planning Workshop (2009)

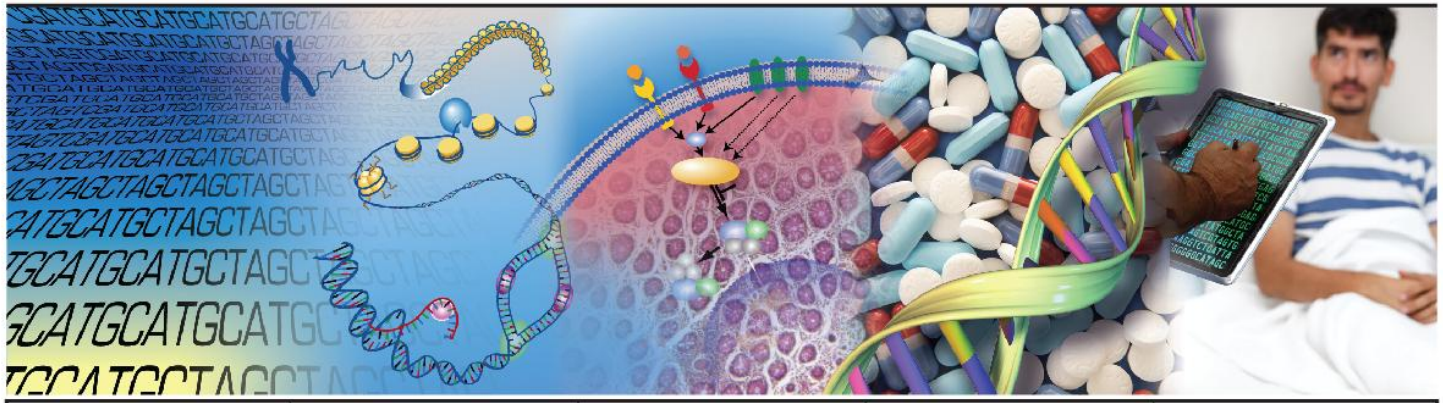
Understanding the Structure of Genomes

Understanding the Biology of Genomes

Understanding the Biology of Disease

Advancing the Science of Medicine

Improving the Effectiveness of Healthcare



1990-2003
Human Genome Project

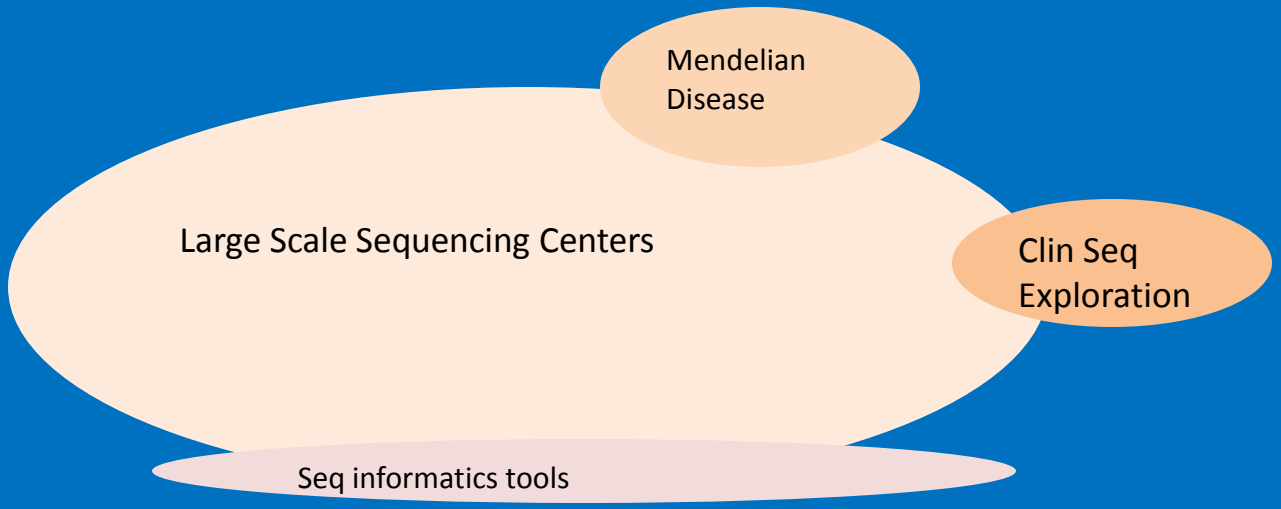
2004-2010

2011-2020

Beyond 2020

Reduce Large Scale Centers, Add Three New Activities

STRUCTURE	BIOLOGY OF GENOMES	BIOLOGY OF DISEASE	SCIENCE OF MEDICINE	IMPR HEALTH CARE

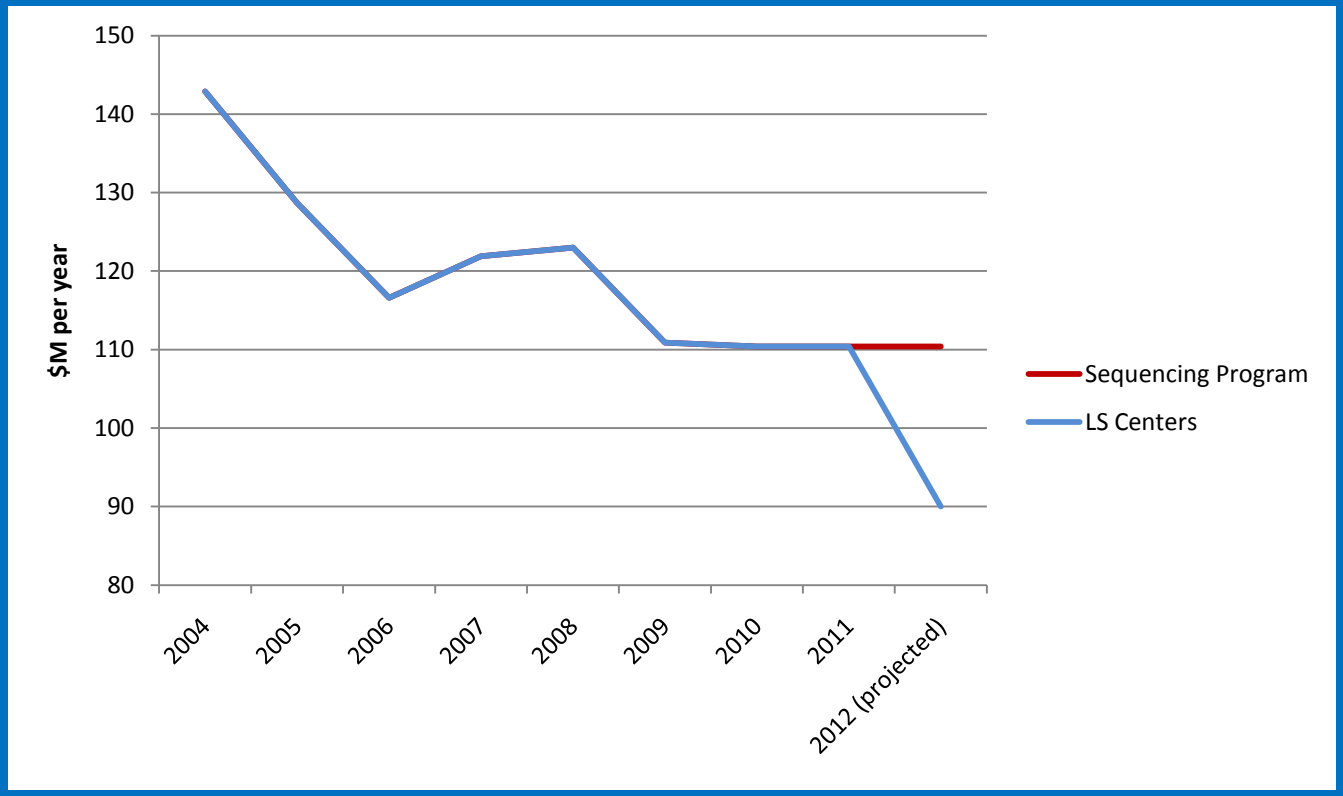


New Sequencing Program: Four RFA's

1. **LS Centers** ($90M/year) – continued benefits of large-scale: flexible, resource development, major projects that require scale or other unique features
2. **Mendelian Centers** ($10 M/year) – focus to organize identification of all variants underlying Mendelian disease
3. **Clinical Sequencing Exploration Centers** ($5.5 M/year) – identify requirements for routine clinical use by "doing".
4. **Sequencing Informatics Tools** ($4 M/year + SBIR) – speed up dissemination of sequencing by encouraging the "robustification" of critical informatics tools

New Funding Picture

Funding ($M/Year)



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